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OM protein - protein search, using sw model

Run on: September 11, 2006, 22:15:38 ; Search time 209.448 Seconds
(without alignments)
4398.662 Million cell updates/sec

Title: US-10-632-342-8
Perfect score: 10489
Sequence: 1 MANFLPRGTSFRRFTRES.....HSEDLADPPSPDRDRESIV 2015

Scoring table: BLOSUM62
Gapop 10.0 , Gapept 0.5

Searched: 2589679 seqs, 457216429 residues

Total number of hits satisfying chosen parameters: 2589679

Minimum DB seq length: 0
Maximum DB seq length: 2000000000

Post-processing: Minimum Match 0%
Maximum Match 100%
Listing first 200 summaries

Database :

A Geneseq_8:.*
1: Geneseqp1980s:.*
2: Geneseqp1990s:.*
3: Geneseqp2000s:.*
4: Geneseqp2001s:.*
5: Geneseqp2002s:.*
6: Geneseqp2003as:.*
7: Geneseqp2003bs:.*
8: Geneseqp2004s:.*
9: Geneseqp2005s:.*
10: Geneseqp2006s:.*

Pred. No. is the number of results predicted by chance to have a score greater than or equal to the score of the result being printed, and is derived by analysis of the total score distribution.

SUMMARIES

Result No.	Score	Query Match	Length	ID	Description
1	10489	100.0	2015	8 ADM34001	Adm34001 Human SCN
2	10487	99.9	2015	7 ADF56441	Adf56441 Human Nav
3	10484	99.9	2015	8 ADM33999	Adm33999 Human SCN
4	10484	99.9	2015	8 AEB22972	Aeb22972 Sodium ch
5	10478.5	99.9	2015	8 ADM33997	Adm33997 Human SCN
6	10478	99.9	2015	9 AEB22993	Aeb22993 Mutant hu
7	10478	99.9	2015	9 AEB22994	Aeb22994 Mutant hu
8	10473.5	99.9	2015	8 ADM33995	Adm33995 Human SCN
9	10473.5	99.9	2015	10 AEF90518	Aef90518 Human SCN
10	10471	99.8	2015	9 AEA78664	Aea78664 Human SCN
11	10432.5	99.5	2016	4 AAB82239	Aab82239 Human SCN
12	10432.5	99.5	2016	7 ADDE4756	Add4756 Human Pro
13	10432.5	99.5	2016	7 ADE55106	Ades5106 Human Pro
14	10429.5	99.4	2016	4 AAB82245	Aab82245 Human SCN
15	10429.5	99.4	2016	4 AAB82241	Aab82241 Human SCN
16	10428.5	99.4	2016	4 AAB82244	Aab82244 Human SCN
17	10427.5	99.4	2016	4 AAB82240	Aab82240 Human SCN
18	10425.5	99.4	2016	4 AAB82243	Aab82243 Human SCN
19	10421.5	99.4	2016	2 AAW23994	Aaw23994 Human htl
20	10416	99.3	2015	4 AAB82242	Aab82242 Human SCN
21	9767	93.1	2019	2 AAR67913	Aar67913 Cardiac s
22	9719	92.7	2020	2 AAR06584	Aar06584 Cardiac s
23	7659.5	73.0	1603	4 AAU19518	Aau19518 Human dia

24	6406.5	61.1	2000	8 ADP79541	Adp79541 Human sod
25	6394.5	61.0	2005	4 AAB99676	Aab99676 Human adu
26	6394.5	61.0	2005	9 ADY27148	Ady27148 Human SCN
27	6393.5	61.0	2005	9 ADY27149	Ady27149 Human SCN
28	6392.5	60.9	2005	5 ABB83627	Abb83627 Human GEf
29	6392.5	60.9	2005	7 ABB78604	Abb78604 Human sod
30	6391.5	60.9	2005	7 ADB78603	Adb78603 Human sod
31	6391.5	60.9	2005	7 ADB78605	Adb78605 Human sod
32	6391.5	60.9	2005	7 ADC46947	Adc46947 Human SCN
33	6391.5	60.9	2005	8 ADS52265	Ads52265 Human bod
34	6391.5	60.9	2005	8 ADY27151	Ady27151 Human SCN
35	6390.5	60.9	2005	4 ADY27150	Ady27150 Human SCN
36	6389.5	60.9	2005	4 AAB99677	Aab99677 Human neo
37	6387.5	60.9	2005	9 ADY27147	Ady27147 Human SCN
38	6377.5	60.8	2000	5 ABB06027	Abb06027 Human bod
39	6377.5	60.8	2000	8 ADK81762	Adk81762 Human Nav
40	6377.5	60.8	2000	8 ADP79545	Adp79545 Human bod
41	6377.5	60.8	2000	9 AEA44248	Aea44248 Human bod
42	6342.5	60.5	1979	4 AEA44245	Aea44245 Murine so
43	6329.5	60.3	2009	4 AAB99674	Aab99674 Human adu
44	6329.5	60.3	2009	7 ABR83180	AbR83180 Human SCN
45	6329.5	60.3	2009	7 ADE57563	Ades7563 Human Pro
46	6327.5	60.3	2009	7 ADE57561	Ades7561 Rat Prote
47	6327.5	60.3	2009	9 ADX26412	Adx26412 Novel cel
48	6324.5	60.3	2009	5 AAE20515	Aae20515 Human ion
49	6324.5	60.3	2009	5 ABB83626	Abb83626 Human GEf
50	6320.5	60.3	2009	8 ADS87515	Ads87515 Mutant SC
51	6319.5	60.2	2009	5 ABB69292	Abb69292 Human bod
52	6319	60.2	1998	7 ABR83184	AbR83184 Human SCN
53	6318	60.2	1998	9 ADX26269	Adx26269 Novel cel
54	6319	60.2	2009	8 AEB43184	Aeb43184 Human pot
55	6317.5	60.2	2009	8 ADS87505	Ads87505 Mutant SC
56	6316.5	60.2	2009	5 ABB69291	Abb69291 Human bod
57	6316.5	60.2	2009	5 ABB69290	Abb69290 Human bod
58	6316.5	60.2	2009	5 AAE16776	Aae16776 Human tra
59	6316.5	60.2	2009	7 ADB78598	Adb78598 Human bod
60	6316.5	60.2	2009	8 ADS87509	Ads87509 Mutant SC
61	6315.5	60.2	2009	8 ADS87513	Ads87513 Mutant SC
62	6315.5	60.2	2009	8 ADS87532	Ads87532 Mutant SC
63	6315.5	60.2	2009	8 ADS87507	Ads87507 Mutant SC
64	6315.5	60.2	2009	7 ADB78593	Adb78593 Human bod
65	6314.5	60.2	2009	8 ADS87508	Ads87508 Mutant SC
66	6314.5	60.2	2009	8 ADS87511	Ads87511 Mutant SC
67	6314.5	60.2	2009	8 ADS87504	Ads87504 Mutant SC
68	6314.5	60.2	2009	8 ADS87530	Ads87530 Mutant SC
69	6314.5	60.2	2009	9 ADS87539	Ads87539 Human SCN
70	6314.5	60.2	1998	5 AAE20510	Aae20510 Human ion
71	6314	60.2	2009	7 ADB78595	Adb78595 Human bod
72	6313.5	60.2	2009	8 ADS87506	Ads87506 Mutant SC
73	6313.5	60.2	2009	9 ADY27142	Ady27142 Human SCN
74	6313.5	60.2	2009	9 ADY27140	Ady27140 Human SCN
75	6313.5	60.2	2009	5 ABB69293	Abb69293 Human bod
76	6312.5	60.2	2009	5 ABB69294	Abb69294 Human bod
77	6312.5	60.2	2009	7 ADB78599	Adb78599 Human bod
78	6312.5	60.2	2009	8 ADS87514	Ads87514 Mutant SC
79	6312.5	60.2	2009	8 ADS87503	Ads87503 Mutant SC
80	6311.5	60.2	2009	5 ABB69289	Abb69289 Human bod
81	6310.5	60.2	2009	8 ADS87502	Ads87502 Mutant SC
82	6310.5	60.2	2009	8 ADS87531	Ads87531 Mutant SC
83	6310.5	60.2	2009	9 ADY27141	Ady27141 Human SCN
84	6310.5	60.2	2009	7 ADB78594	Adb78594 Human bod
85	6309.5	60.2	2009	7 ADB78607	Adb78607 Human bod
86	6308.5	60.1	1950	8 ADP79543	Adp79543 Human bod
87	6308	60.1	1951	8 ADS87510	Ads87510 Mutant SC
88	6307.5	60.1	2009	5 ABB06026	Abb06026 Human bod
89	6300.5	60.1	1999	4 ADU78366	Adu78366 Human vol
90	6300.5	60.0	1999	4 AAB99678	Aab99678 Human adu
91	6289	60.0	1951	5 AAE20516	Aae20516 Human ion
92	6287	59.9	1973	5 ABR83185	AbR83185 Human SCN
93	6285.5	59.9	1981	4 AAB99679	Aab99679 Human neo
94	6283	59.9	1951	8 ADL06576	Adl06576 Human tum
95	6281	59.9	1951	9 AEA44250	Aea44250 Human SCN
96	6281	59.9	1951		

97	6276.5	59.8	1962	5	AAE05011	Human	Ion
98	6272.5	59.8	1961	7	ADBE9628	Rat	Prote
99	6272.5	59.8	1951	7	AE5A4232	Rat	Sodiu
100	6257.5	59.7	1956	4	AA6E5785	Human	SNS
101	6257.5	59.7	1956	4	AA6E1996	Human	per
102	6257.5	59.7	1956	6	AB976193	Human	per
103	6257.5	59.7	1956	6	AB976193	Human	per
104	6257.5	59.7	1956	6	ADAs0152	Human	per
105	6257.5	59.7	1956	6	ADAs0152	Human	per
106	6257.5	59.7	1956	8	ADUf08471	Human	heea
107	6257.5	59.7	1956	8	ADUf08471	Human	Nav
108	6257.5	59.7	1956	8	ADUf11150	Human	sod
109	6257.5	59.7	1956	8	ADUf11150	Human	sod
110	6253.5	59.6	1956	8	ADU71152	Human	per
111	6251.5	59.6	1956	6	AB975945	Human	per
112	6245	59.5	1954	7	ADBE45451	Rat	Prote
113	6245	59.5	1954	7	ADBE45451	Rat	Prote
114	6245	59.5	1954	7	ADBE33027	Rat	Prote
115	6213.5	59.2	1977	8	ADU73202	Human	per
116	6213.5	59.2	1977	8	ADU73202	Human	per
117	6213.5	59.2	1977	9	ADBE66379	Human	per
118	6213.5	59.2	1978	7	ADBE64553	Human	Pro
119	6213.5	59.2	1978	7	ADBE45459	Human	Pro
120	6212.5	59.2	1977	9	AE8B6346	Amino	aci
121	6212.5	59.2	1977	9	AE8B6347	Amino	aci
122	6212.5	59.2	1977	9	AE8B6343	Amino	aci
123	6212.5	59.2	1977	9	AE8B6348	Amino	aci
124	6210.5	59.2	1977	9	AE8B6345	Amino	aci
125	6207.5	59.2	1977	2	AA939641	Periphera	
126	6206	59.2	1978	2	AA969361	Tetradoc	
127	6205.5	59.2	1977	2	AE8B6344	Amino	aci
128	6198	59.1	1978	9	ADX66338	Novel	cel
129	6197	59.1	1980	8	ADZ88372	Human	SCN
130	6197	59.1	1980	2	AA9E9332	Tetradoc	
131	6194	59.1	1980	2	ADB78600	Human	sod
132	6194	59.1	1984	7	AA939639	Periphera	
133	6189	59.0	1980	7	ADB78605	Human	sod
134	6186	59.0	1989	2	AA932317	Periphera	
135	6178	58.9	1942	8	ADBS87534	Mutant	SC
136	6140	58.5	1980	3	AA933563	Human	sod
137	6140	58.5	1980	5	AA014927	Human	sod
138	6138.5	58.5	1962	2	AA917250	NANG	poly
139	6110.5	58.2	1942	8	ADUJ1164	Guinea	pi
140	6103.5	58.2	1957	6	ADAs0156	Rat	perip
141	6100	58.2	1956	4	AA9E5783	Rat	SNS1
142	6100	58.2	1956	4	AA9E1995	Rat	perip
143	6100	58.2	1956	6	ADAs0153	Rat	perip
144	6100	58.2	1956	6	ADUJ1165	Rat	sodiu
145	6100	58.2	1956	9	AE8B90788	Rat	perip
146	6088.5	58.0	1957	2	AA9W1740	Variant	r
147	6087.5	58.0	1957	6	AB976191	Rat	voluta
148	6087.5	58.0	1957	7	ADDA4754	Rat	Prote
149	6087.5	58.0	1957	8	ADFP08469	Rat	Nav
150	6087.5	58.0	1957	8	ADUJ1148	Rat	sodiu
151	6087.5	58.0	1957	9	ADX66336	Novel	cel
152	6086.5	58.0	1957	2	AA9W1737	Wild type	
153	6078	57.9	1891	9	ADY72145	Rat	perip
154	6078	57.9	1958	4	AA9E5784	Mouse	SNS
155	6078	57.9	1958	4	AA9E5784	Mouse	SNS
156	6066.5	57.8	1836	7	ADBE59630	Human	Pro
157	6066.5	57.8	1836	7	ADBE59630	Human	Pro
158	6066.5	57.8	1836	7	ADBE59630	Human	Pro
159	6060.5	57.8	1836	6	ADQ17412	Human	sof
160	6059.5	57.8	1836	9	AEF53825	Human	vol
161	6049.5	57.7	1957	6	ADAs0155	Rat	perip
162	6049	57.7	1976	7	ADBE57386	Rat	Prote
163	6046	57.6	1956	6	AB975944	Rat	perip
164	6046	57.6	1956	6	ADAs0144	Rat	perip
165	6046	57.6	1956	9	AE8B90778	Rat	perip
166	6043	57.6	2132	2	AA9W1739	Variant	r
167	6012	57.3	1989	2	AA939640	Periphera	
168	5876	56.0	1855	7	ADB78597	Human	sod
169	5644	53.8	1795	7	ADB78596	Human	sod

170	5566.5	53.1	1835	2	AAE23316	Peripheral
171	5455	52.0	1107	6	ABR41495	Human DIR
172	5293	50.5	1276	9	ADZ86370	Human SCN
173	4811	45.9	1765	9	AAI06537	Mouse boot
174	4811	45.9	1765	4	AAE20124	Mouse boot
175	4811 ⁴	45.9	1765	7	ADD32186	Mouse Na
176	4811	45.9	1765	9	ADZ26317	Novel cel
177	4729	45.1	1791	5	AAI04925	AAI04925 Human cel
178	4729	45.1	1791	9	ADZ6245	Novel cel
179	4729	45.1	1825	9	AED97979	Human SCN
180	4728	45.1	1765	7	ADD32182	Rat Na v
181	4728	45.1	1765	7	ADD32210	Rat Na v
182	4728	45.1	1765	5	ADDE5104	Rat Prote
183	4728	45.1	1765	9	ADZ63839	Novel cel
184	4724	45.0	1765	2	AAI16572	Type 5 sc
185	4724	45.0	1791	4	AAE20121	Human foot
186	4724	45.0	1791	7	ADD32194	Human Na
187	4720	45.0	1765	2	AAI06556	Rat sodiu
188	4720	45.0	1765	4	AAE20122	Rat sodiu
189	4720	45.0	1765	4	AAE20123	Rat sodiu
190	4719	45.0	1765	2	AAI1668	Rat sense
191	4711	44.9	1753	9	AED97979	Human SCN
192	4488.5	42.8	1510	8	ADZ87522	Mutant SC
193	4207.5	40.1	1453	5	AAE20517	Human ion
194	4197	39.8	1442	5	AAE20512	Human ion
195	4173.5	39.8	2131	4	ABE40743	Drosophil
196	4173.5	39.8	2131	8	ADJ30141	Drosophil
197	4169.5	39.8	2131	8	ADJ30146	Drosophil
198	4167	39.7	2105	2	AAE57772	Mouse dom
199	4167	39.7	2105	2	AAE59577	Calcium F
200	4162	39.7	2104	2	AAW57773	Mouse dom

ALIGNMENTS

RESULT 1
ADM34001

DT	03-JUN-2004	(first entry)
XX	Human SCN5A variant 4	protein SEQ ID NO:8
DE		

KM human; cardiac sodium channel alpha subunit; SCN5A;
KM sodium voltage-gated channel; type V; alpha; mutation study
KM sodium channel related disease.

OS Homo sapiens.

PN WO2004012668-A2.

PD 12-FEB-2004.

PF 01-AUG-2003; 2003WO-US024190

PR 02-AUG-2002; 2002US-0401018P

PA (WISC) WISCONSIN ALUMNI RES FOUND.

PI Makielski JC, Ye B, Ackerman MJ;

DR WPI; 2004-157000/15.

XX

PT polypeptide variants, useful for studying mutations, and designing or

PT channel related diseases.

PS Claim 17; SEQ ID NO 8; 111pp; English.

XX The present sequence represents a variant of the human cardiac sodium channel alpha subunit designated SCN5A (sodium voltage-gated channel, type V, alpha) protein. The present invention describes an isolated polynucleotide encoding an SCN5A polypeptide where the polynucleotide is selected from the group: (1) a first polynucleotide that encodes an SCN5A polypeptide selected from the group consisting of: (i) a histidine, threonine, leucine, arginine and glutamine at amino acid positions 558, 559, 618, 1027 and 1077, respectively; (ii) an arginine, threonine, leucine, arginine and glutamine at amino acid positions 558, 559, 618, 1027 and 1077, respectively; (iii) an arginine, threonine, leucine, arginine and glutamine at amino acid positions 558, 559, 618, 1027 and 1077, respectively; (iv) an arginine, threonine, leucine, arginine and glutamine at amino acid positions 558, 559, 618 and 1027, respectively, with the amino acid at amino acid position 1077 deleted; (2) a second polynucleotide that is at least 80 % identical to the first polynucleotide over the entire length of the first polynucleotide; (3) a third polynucleotide that encodes any of the SCN5A polypeptides with a conservative substitution, deletion or rearrangement at one or more non-critical amino acid position; and (4) a fourth polynucleotide that is a complement of the first, second or third polynucleotide. Also described: (1) a genetic construct comprising the polynucleotide operably linked to a non-native expression control sequence; (2) a cell comprising the polynucleotide; (3) an isolated polypeptide encoded by the polynucleotide; (4) an antibody that specifically binds to the polypeptide; (5) identifying an agent that can alter the activity of a sodium channel; (6) identifying an agent that can alter the expression of a sodium channel; (7) determining whether a biological sample or a preparation derived from the biological sample contains the polypeptide; (8) determining whether a mutation on a sodium channel is associated with a disease; and (9) determining whether a human or non-human subject is at risk for long QT syndrome. The SCN5A polynucleotides and polypeptides are useful for studying mutations, and designing or identifying new diagnostics and treatment strategies or agents for sodium channel related diseases or conditions.

Sequence 2015 AA;

Query Match 100.0%; Score 10489; DB 8; Length 2015;
Beet Local Similarity 100.0%; Pred. No. 0;
Matches 2015; Conservative 0; Mismatches 0; Indels 0; Gaps 0;

QY 1 MANFLPRGTSFRRFRESLAIAEKMAEKQARGSTTQESREGPEEAREPQLODOA 60
DB 1 MANFLPRGTSFRRFRESLAIAEKMAEKQARGSTTQESREGPEEAREPQLODOA 60
QY 61 SKGLPDLVGNPQELIGEBLEDLPFYSTQKTFIVLNGKTIFFRSATNALVLSPPHPI 120
DB 61 SKGLPDLVGNPQELIGEBLEDLPFYSTQKTFIVLNGKTIFFRSATNALVLSPPHPI 120
QY 121 RRAVKILVHSLFNNLIMCTITLNCVMAQHDPPMTKYVEYTFALITFESLVKILARG 180
DB 121 RRAVKILVHSLFNNLIMCTITLNCVMAQHDPPMTKYVEYTFALITFESLVKILARG 180
QY 181 PCLHAFTLRDPMNLDFSIIIMATTEFVDLGNVSAIRTPFVLAALKTISTISGLKTIY 240
DB 181 PCLHAFTLRDPMNLDFSIIIMATTEFVDLGNVSAIRTPFVLAALKTISTISGLKTIY 240
QY 241 GALIGSVKKADVWVLTVCISVFPALIGLOFMGNLRHKCVANFTALNGTNGSVEADGLV 300
DB 241 GALIGSVKKADVWVLTVCISVFPALIGLOFMGNLRHKCVANFTALNGTNGSVEADGLV 300
QY 301 WESLDLYISDPENYLLKNGTSVYLLCGNSSDAGTCEGRCCLKAGENDHGTSTDSFAM 360
DB 301 WESLDLYISDPENYLLKNGTSVYLLCGNSSDAGTCEGRCCLKAGENDHGTSTDSFAM 360
QY 361 AFLALFRLMTQDCWRLYQOTLRSAKTIYMFMLVIFLGSFYLVNLLAVANAYEEN 420
DB 361 AFLALFRLMTQDCWRLYQOTLRSAKTIYMFMLVIFLGSFYLVNLLAVANAYEEN 420
QY 421 QATIAETEKEKRRFOEAMEMLKKEHEALTIRGVDTVSSRSSLEMSPLAPVNSHERSKRK 480
DB 421 QATIAETEKEKRRFOEAMEMLKKEHEALTIRGVDTVSSRSSLEMSPLAPVNSHERSKRK 480

QY 481 RMSSGTECEGDRLPKSDSEDPARAMNHLSTRGLSRTSMKRRSGSIFFTRRRDLGSE 540
DB 481 RMSSGTECEGDRLPKSDSEDPARAMNHLSTRGLSRTSMKRRSGSIFFTRRRDLGSE 540
QY 541 ADPADENSTAGESSHRTSLVWPLRRTSAQGPSPTGAPGALHNGKNSYVDCNGV 600
DB 541 ADPADENSTAGESSHRTSLVWPLRRTSAQGPSPTGAPGALHNGKNSYVDCNGV 600
QY 601 VSLGADPBEATSPGSHLRPVMLEHPPTTTPSEPGPGQWLTQAPCVQDFEEGAAQ 660
DB 601 VSLGADPBEATSPGSHLRPVMLEHPPTTTPSEPGPGQWLTQAPCVQDFEEGAAQ 660
QY 661 RALSASVLTSLAELESBRHKCPQCMRLAORYLIMECCPLMMSIKQGVKLVWMDPFD 720
DB 661 RALSASVLTSLAELESBRHKCPQCMRLAORYLIMECCPLMMSIKQGVKLVWMDPFD 720
QY 721 LITTCIVLNTLFMALHEHYNMTSFEEMLOVGNLFTFTGIFTAEMTFKLTALDPYFFQOG 780
DB 721 LITTCIVLNTLFMALHEHYNMTSFEEMLOVGNLFTFTGIFTAEMTFKLTALDPYFFQOG 780
QY 781 WNIPIISIIIVLSLMEGLSRMSNLSVLRSPRLRVFKLAKSWPTLNTLKIIGNSVGALG 840
DB 781 WNIPIISIIIVLSLMEGLSRMSNLSVLRSPRLRVFKLAKSWPTLNTLKIIGNSVGALG 840
QY 841 NLTIVLATTIIFFAVVGMOLOGKNSYSELSDSGLLPMMHMDPFAHPLITRIICGEMI 900
DB 841 NLTIVLATTIIFFAVVGMOLOGKNSYSELSDSGLLPMMHMDPFAHPLITRIICGEMI 900
QY 901 ETMDMCEVSGQSLCLVFLVMVIGNLVNLFPLALISFSADNLTAPADEREANNLQ 960
DB 901 ETMDMCEVSGQSLCLVFLVMVIGNLVNLFPLALISFSADNLTAPADEREANNLQ 960
QY 961 LALARIQGLRFRKRTTWDPCGGLRQRPQKRALAAGQULPSCIATPYSPPEETKXP 1020
DB 961 LALARIQGLRFRKRTTWDPCGGLRQRPQKRALAAGQULPSCIATPYSPPEETKXP 1020
QY 1021 PTRKREBEBOPOGPRPDPEPCVPIAAVSEPTDQOEDEENSLGTEESSSQOESOP 1080
DB 1021 PTRKREBEBOPOGPRPDPEPCVPIAAVSEPTDQOEDEENSLGTEESSSQOESOP 1080
QY 1081 VSGGEAPDPSRTMSQVATASSEAEASQADNMQKAEPOAGCETPEDESCEGST 1140
DB 1081 VSGGEAPDPSRTMSQVATASSEAEASQADNMQKAEPOAGCETPEDESCEGST 1140
QY 1141 ADMNTAELBQIPDLGQDVXDPBDCFTGECVRRCPCCAVDTTQAPGKVMWRLRTCYHI 1200
DB 1141 ADMNTAELBQIPDLGQDVXDPBDCFTGECVRRCPCCAVDTTQAPGKVMWRLRTCYHI 1200
QY 1201 VEHSWFEFTIIFMILSSGALAFEDIYLBKRTIKVLEAYDKMTTYFVLEMLKWAY 1260
DB 1201 VEHSWFEFTIIFMILSSGALAFEDIYLBKRTIKVLEAYDKMTTYFVLEMLKWAY 1260
QY 1261 GFKKYFTYAMCMLPDLVDVSLVANTLFAEWGPIKSLRTALAPLRLASFEGMR 1320
DB 1261 GFKKYFTYAMCMLPDLVDVSLVANTLFAEWGPIKSLRTALAPLRLASFEGMR 1320
QY 1321 VVNAALVGAISIMNVLLVCLIFMLIPSIMGVNLPAKRGRCINOTEGDPLNNTIYNNK 1380
DB 1321 VVNAALVGAISIMNVLLVCLIFMLIPSIMGVNLPAKRGRCINOTEGDPLNNTIYNNK 1380
QY 1381 SOCESLNTLGFLLYTKVNFNDNAGAGTALLQVATFFGMMDDIWAADAUSGIEBOPOWE 1440
DB 1381 SOCESLNTLGFLLYTKVNFNDNAGAGTALLQVATFFGMMDDIWAADAUSGIEBOPOWE 1440
QY 1441 YNLWYIYFVFIIFGSPFTNLFPAGVINDFNQOKKLGODIFMTEQOKKYNYAMKGL 1500
DB 1441 YNLWYIYFVFIIFGSPFTNLFPAGVINDFNQOKKLGODIFMTEQOKKYNYAMKGL 1500
QY 1501 GSKRPQKPIPRPLNKYQGFIDYVTKQAFDVTIMFLCLNNVTMAVETDDQSPKXINILA 1560
DB 1501 GSKRPQKPIPRPLNKYQGFIDYVTKQAFDVTIMFLCLNNVTMAVETDDQSPKXINILA 1560

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Qy 1561 KINLLPVAIFPGECLVLAALRHYFTNNSWNIPEVWVLIISVGTVLSDIIQKXFESEPTL 1620
Cc 1561 KINLLPVAIFPGECLVLAALRHYFTNNSWNIPEVWVLIISVGTVLSDIIQKXFESEPTL 1620
Db 1561 KINLLPVAIFPGECLVLAALRHYFTNNSWNIPEVWVLIISVGTVLSDIIQKXFESEPTL 1620
Qy 1621 FVIRLRLRIGILRLIRGAKGIRTLTFLMMSLPALFENIGLLFLVMEFYSIFGMANFAY 1680
Cc 1621 FVIRLRLRIGILRLIRGAKGIRTLTFLMMSLPALFENIGLLFLVMEFYSIFGMANFAY 1680
Db 1621 FVIRLRLRIGILRLIRGAKGIRTLTFLMMSLPALFENIGLLFLVMEFYSIFGMANFAY 1680
Qy 1681 VKMEAGIDMFNFQTFANSMCLFQITTSAGMDGLSPILNTGPPYCDPLPNSNGSRGD 1740
Cc 1681 VKMEAGIDMFNFQTFANSMCLFQITTSAGMDGLSPILNTGPPYCDPLPNSNGSRGD 1740
Db 1681 VKMEAGIDMFNFQTFANSMCLFQITTSAGMDGLSPILNTGPPYCDPLPNSNGSRGD 1740
Qy 1741 CGSPAVGLLEFTTYIIISFLVWVNYIAIILENSVATEESTEPSEDDPFMEIWEKF 1800
Cc 1741 CGSPAVGLLEFTTYIIISFLVWVNYIAIILENSVATEESTEPSEDDPFMEIWEKF 1800
Db 1741 CGSPAVGLLEFTTYIIISFLVWVNYIAIILENSVATEESTEPSEDDPFMEIWEKF 1800
Qy 1801 DPEATQFIEYSVLSPFADALSEPRLAKPNOISLINMDLPVWSGDRICMIDILFAFTKRV 1860
Cc 1801 DPEATQFIEYSVLSPFADALSEPRLAKPNOISLINMDLPVWSGDRICMIDILFAFTKRV 1860
Db 1801 DPEATQFIEYSVLSPFADALSEPRLAKPNOISLINMDLPVWSGDRICMIDILFAFTKRV 1860
Qy 1861 LGESGEMDALKIOMEKEFMAANPSKISYEPIITTLRRGHEVSAMVIOQAFRRHLQRL 1920
Cc 1861 LGESGEMDALKIOMEKEFMAANPSKISYEPIITTLRRGHEVSAMVIOQAFRRHLQRL 1920
Db 1861 LGESGEMDALKIOMEKEFMAANPSKISYEPIITTLRRGHEVSAMVIOQAFRRHLQRL 1920
Qy 1921 KHAFTLFRQOAGSGISEEDAPEREGLIAYVMSSENSRPLGPPSSSISTSPPSYDSVT 1980
Cc 1921 KHAFTLFRQOAGSGISEEDAPEREGLIAYVMSSENSRPLGPPSSSISTSPPSYDSVT 1980
Db 1921 KHAFTLFRQOAGSGISEEDAPEREGLIAYVMSSENSRPLGPPSSSISTSPPSYDSVT 1980
Qy 1981 RATSNDLQVRGSDYSHSEDLADFPSPDRDRESIV 2015
Cc 1981 RATSNDLQVRGSDYSHSEDLADFPSPDRDRESIV 2015
Db 1981 RATSNDLQVRGSDYSHSEDLADFPSPDRDRESIV 2015

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RESULT 2
ADFS6441
ID ADFS6441 standard, protein; 2015 AA.

AC ADFS6441;

DT 12-FEB-2004 (first entry)

XX Human Nav1.5 sodium channel alpha subunit SCN5A h1b.

XX cardiac; antiarrhythmic; sodium channel blocker;

XX sodium channel activator; SCN5A channel blocker; SCN5A channel activator;

XX sodium channel subunit h1b; h1b; SCN5A; Long QT syndrome;

XX sodium channel; cardiac arrhythmia; heart disorder;

XX human Nav1.5 sodium channel alpha subunit SCN5A.

XX Homo sapiens.

XX US2003157600-A1.

XX 21-AUG-2003.

XX 12-FEB-2002; 2002US-00077054.

XX 12-FEB-2002; 2002US-00077054.

XX (MAKI/) MAKIELSKI J C.

XX (YEBB/) YE B.

XX Makielecki JC, Ye B;

XX WPI; 2003-688984/65.

XX N-PESDB; ADFS6440.

XX Claim 1, SEQ ID NO 2; 24pp; English.

CC The invention describes an isolated sodium channel subunit h1b
CC polypeptide (I) comprising a fully defined 2015 amino acid sequence as
CC given in the specification optionally, carrying a conservative
CC substitution, deletion or rearrangement at one or more non-critical amino
CC acid position. Detecting the presence of the h1b SCN5A gene is useful
CC for determining if a (non-)human subject is at risk for Long QT syndrome.
CC Agents screened that increase or decrease sodium channel activity are
CC useful for treating cardiac arrhythmia and other sodium channel related
CC heart disorders. This is the amino acid sequence of human Nav1.5 sodium
CC channel alpha subunit SCN5A.

Sequence 2015 AA;
Query Match 4 99.9%; Score 10487; DB 7; Length 2015;
Best Local Similarity 99.9%; Pred. No. 0;
Matches 2014; Conservative 1; Mismatches 0; Indels 0; Gaps 0;

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Qy 1 MANFLPRTGSSPRFRRESIAAIEKMAEKQARGSTTLQESREGLEPEEAPRQDLQA 60
Cc 1 MANFLPRTGSSPRFRRESIAAIEKMAEKQARGSTTLQESREGLEPEEAPRQDLQA 60
Db 1 MANFLPRTGSSPRFRRESIAAIEKMAEKQARGSTTLQESREGLEPEEAPRQDLQA 60
Qy 61 SKKLPLDYGNPPOELICEPLEDDPPFYSTQKTIIVLNKGTIRFSAITNLVYLSPPHP 120
Cc 61 SKKLPLDYGNPPOELICEPLEDDPPFYSTQKTIIVLNKGTIRFSAITNLVYLSPPHP 120
Db 61 SKKLPLDYGNPPOELICEPLEDDPPFYSTQKTIIVLNKGTIRFSAITNLVYLSPPHP 120
Qy 121 RRAAVKILVHSLFVNLIMCTILNVCVMAQHDPPPTKYEYTFATYFESLVKILARG 180
Cc 121 RRAAVKILVHSLFVNLIMCTILNVCVMAQHDPPPTKYEYTFATYFESLVKILARG 180
Db 121 RRAAVKILVHSLFVNLIMCTILNVCVMAQHDPPPTKYEYTFATYFESLVKILARG 180
Qy 181 FCLHAFPLNDPNNWMLDFSVIIMAYTTEFVLDGNSALRTFVRLAKTISVIGLKTIV 240
Cc 181 FCLHAFPLNDPNNWMLDFSVIIMAYTTEFVLDGNSALRTFVRLAKTISVIGLKTIV 240
Db 181 FCLHAFPLNDPNNWMLDFSVIIMAYTTEFVLDGNSALRTFVRLAKTISVIGLKTIV 240
Qy 241 GALIQSVKCLADVWVLTFFCLSVFALIGLQFPKMLRHKCVNPTALNGNSVEADGLV 300
Cc 241 GALIQSVKCLADVWVLTFFCLSVFALIGLQFPKMLRHKCVNPTALNGNSVEADGLV 300
Db 241 GALIQSVKCLADVWVLTFFCLSVFALIGLQFPKMLRHKCVNPTALNGNSVEADGLV 300
Qy 301 WESLDLYLDPENYLNKGTSDVLLCGNSSDAGCPREGYCLXAGENPDHGYTSFDSFAW 360
Cc 301 WESLDLYLDPENYLNKGTSDVLLCGNSSDAGCPREGYCLXAGENPDHGYTSFDSFAW 360
Db 301 WESLDLYLDPENYLNKGTSDVLLCGNSSDAGCPREGYCLXAGENPDHGYTSFDSFAW 360
Qy 361 AFLALFRLMTQDCWERLYQOTLRGAKIYIMFEMLVIFICSFYLVNLLAVVMAAYEON 420
Cc 361 AFLALFRLMTQDCWERLYQOTLRGAKIYIMFEMLVIFICSFYLVNLLAVVMAAYEON 420
Db 361 AFLALFRLMTQDCWERLYQOTLRGAKIYIMFEMLVIFICSFYLVNLLAVVMAAYEON 420
Qy 421 QATTAETBEKRRQGEAMWELKKEHEALTTRGVDYRSLSLEWPLAPVNSHERSRK 480
Cc 421 QATTAETBEKRRQGEAMWELKKEHEALTTRGVDYRSLSLEWPLAPVNSHERSRK 480
Db 421 QATTAETBEKRRQGEAMWELKKEHEALTTRGVDYRSLSLEWPLAPVNSHERSRK 480
Qy 481 RMSGTECEGEDRLPKSDSEDPNANMLSLTRGLSRSMKPRSSRSISFTFRRDIGSE 540
Cc 481 RMSGTECEGEDRLPKSDSEDPNANMLSLTRGLSRSMKPRSSRSISFTFRRDIGSE 540
Db 481 RMSGTECEGEDRLPKSDSEDPNANMLSLTRGLSRSMKPRSSRSISFTFRRDIGSE 540
Qy 541 ADPADENSTAGESESHRTSLVWPPLRRTSAQCPGTSAPGHALHGXKNSTVDCNV 600
Cc 541 ADPADENSTAGESESHRTSLVWPPLRRTSAQCPGTSAPGHALHGXKNSTVDCNV 600
Db 541 ADPADENSTAGESESHRTSLVWPPLRRTSAQCPGTSAPGHALHGXKNSTVDCNV 600
Qy 601 VSLIAGDPPEATSPGSHILRPWMLERPDPTTPEEBEGGQMLTSQAPCVDGEFEERARQ 660
Cc 601 VSLIAGDPPEATSPGSHILRPWMLERPDPTTPEEBEGGQMLTSQAPCVDGEFEERARQ 660
Db 601 VSLIAGDPPEATSPGSHILRPWMLERPDPTTPEEBEGGQMLTSQAPCVDGEFEERARQ 660
Qy 661 RALSAVSVLTSALTELESRRKCPCCNRLAQRILIMECCPLMMSIQGVKLVMDPFTD 720
Cc 661 RALSAVSVLTSALTELESRRKCPCCNRLAQRILIMECCPLMMSIQGVKLVMDPFTD 720
Db 661 RALSAVSVLTSALTELESRRKCPCCNRLAQRILIMECCPLMMSIQGVKLVMDPFTD 720
Qy 721 LITMCIVLNTLFWALBHYNNTSEFEMLOVGNLVFTGIFTAEMTFKIALDPYYRQOG 780
Cc 721 LITMCIVLNTLFWALBHYNNTSEFEMLOVGNLVFTGIFTAEMTFKIALDPYYRQOG 780
Db 721 LITMCIVLNTLFWALBHYNNTSEFEMLOVGNLVFTGIFTAEMTFKIALDPYYRQOG 780
Qy 781 WNIFDSIIIVILSMELGLSRMSNTSVLRSPFLRLAVFKLAKSWPTLNTLIKIGSVGALG 840
Cc 781 WNIFDSIIIVILSMELGLSRMSNTSVLRSPFLRLAVFKLAKSWPTLNTLIKIGSVGALG 840
Db 781 WNIFDSIIIVILSMELGLSRMSNTSVLRSPFLRLAVFKLAKSWPTLNTLIKIGSVGALG 840

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Qy 841 NLTVALIIVFI FAVVGMQLFGKNYSELSDSGLLPRWMMDFFAFIIRILGEMI 900
    |||
Db 841 NLTVALIIVFI FAVVGMQLFGKNYSELSDSGLLPRWMMDFFAFIIRILGEMI 900
Qy 901 ETTMDCMEVSGSLCLVFLVWVIGNLVNLFLALLSSFSADNLTA PDEDEMNLIQ 960
    |||
Db 901 ETTMDCMEVSGSLCLVFLVWVIGNLVNLFLALLSSFSADNLTA PDEDEMNLIQ 960
Qy 961 LALARIQRGLRFRVKTPTMDFCCGLLRQRQKPAALAAOQLRSCITAPYSPPEPEKXP 1020
    |||
Db 961 LALARIQRGLRFRVKTPTMDFCCGLLRQRQKPAALAAOQLRSCITAPYSPPEPEKXP 1020
Qy 1021 PTKETRFEEGEPGCGTGPDEPVCVPIAVAESDTPDEBEBENSLGTEEBESSKQESOP 1080
    |||
Db 1021 PTKETRFEEGEPGCGTGPDEPVCVPIAVAESDTPDEBEBENSLGTEEBESSKQESOP 1080
Qy 1081 VSGGEPAPDSRTWGSVATASSEAEASASQADMROQWABPOAPCGETPEDSCSGEST 1140
    |||
Db 1081 VSGGEPAPDSRTWGSVATASSEAEASASQADMROQWABPOAPCGETPEDSCSGEST 1140
Qy 1141 ADMTTAELEBQIPDLGQDVKPEDCFTGCTRCGCCAVDTTQAPGKVMRLKRTCYHI 1200
    |||
Db 1141 ADMTTAELEBQIPDLGQDVKPEDCFTGCTRCGCCAVDTTQAPGKVMRLKRTCYHI 1200
Qy 1201 VEHSMFEFPIIFMILSSGALAFBDYLBERTIKVLEADKMFTYVFLMILKVMVAY 1260
    |||
Db 1201 VEHSMFEFPIIFMILSSGALAFBDYLBERTIKVLEADKMFTYVFLMILKVMVAY 1260
Qy 1261 GFKKFTTNAMCWLDFLVDVSLVSVANTLGAEMGPISKLRTLRALRALSFEQNR 1320
    |||
Db 1261 GFKKFTTNAMCWLDFLVDVSLVSVANTLGAEMGPISKLRTLRALRALSFEQNR 1320
Qy 1321 VVWVALVGAIPBSINAVLVCLIFMLIFSIIMGVNLPAGKRGRCINOTEGDPLANTYIVNKK 1380
    |||
Db 1321 VVWVALVGAIPBSINAVLVCLIFMLIFSIIMGVNLPAGKRGRCINOTEGDPLANTYIVNKK 1380
Qy 1381 SOCESLNTGELVMTKVKVNPNGVGYALLQVATPFKMMIMYAAVDSRGYEBOPWE 1440
    |||
Db 1381 SOCESLNTGELVMTKVKVNPNGVGYALLQVATPFKMMIMYAAVDSRGYEBOPWE 1440
Qy 1441 YNLVWYIVFVFIIFGSPFTLNLFTGVLIIDNFOOKKLGQODIMTEBOKKYANAMKL 1500
    |||
Db 1441 YNLVWYIVFVFIIFGSPFTLNLFTGVLIIDNFOOKKLGQODIMTEBOKKYANAMKL 1500
Qy 1501 GSKKQOKPIPRPIANKYOGFIPIVTKQAFDVTIMELCLINMTVMWVEDTDQSPERKINILA 1560
    |||
Db 1501 GSKKQOKPIPRPIANKYOGFIPIVTKQAFDVTIMELCLINMTVMWVEDTDQSPERKINILA 1560
Qy 1561 KINLFFVAIFGECIVKLAALRHXYFTNSWNI FDFVVVLTSLVGTVLSDIIOKXFPSPTL 1620
    |||
Db 1561 KINLFFVAIFGECIVKLAALRHXYFTNSWNI FDFVVVLTSLVGTVLSDIIOKXFPSPTL 1620
Qy 1621 FRVIRIARIGRILIRIGAKGIRITLFPALMNSLPALFNIGLLFTVMFYISFGANAPAY 1680
    |||
Db 1621 FRVIRIARIGRILIRIGAKGIRITLFPALMNSLPALFNIGLLFTVMFYISFGANAPAY 1680
Qy 1661 VKMEAGIDDMENFOFPANSMCLFOITTSAGWDGLSLPINTGPPYCPPTLPNSNGSGRD 1740
    |||
Db 1661 VKMEAGIDDMENFOFPANSMCLFOITTSAGWDGLSLPINTGPPYCPPTLPNSNGSGRD 1740
Qy 1741 CGSPAVGLLFTTYIISPLIVVWNYIAIILENFVATEBESREPSDDPDMFYIWKRF 1800
    |||
Db 1741 CGSPAVGLLFTTYIISPLIVVWNYIAIILENFVATEBESREPSDDPDMFYIWKRF 1800
Qy 1801 DPEATQFLEYSVLSDPADALSEPLRIAKPNQISLINMDLPWVSGDRIRHQMOLFAFTRKV 1860
    |||
Db 1801 DPEATQFLEYSVLSDPADALSEPLRIAKPNQISLINMDLPWVSGDRIRHQMOLFAFTRKV 1860
Qy 1861 LGESGEMDALKIOMEKEMANPSKISYEPIITTLRRHGEVSANVIOAFRRHLLQSSL 1920
    |||
Db 1861 LGESGEMDALKIOMEKEMANPSKISYEPIITTLRRHGEVSANVIOAFRRHLLQSSL 1920
Qy 1921 KHASFLPQOAGSGISEDAPEREGLIAVWSENSENRLGPPSSSISSTSPPSYDSVT 1980
    |||

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Db 1921 KHASFLPQOAGSGISEDAPEREGLIAVWSENSENRLGPPSSSISSTSPPSYDSVT 1980
    |||
Qy 1981 RATSNDLVQVSGSDYSHSEDLADFPSPSPDRRESIV 2015
    |||
Db 1981 RATSNDLVQVSGSDYSHSEDLADFPSPSPDRRESIV 2015
    |||

RESULT 3
ADM3399
ID ADM3399 standard; protein; 2015 AA.
XX
AC ADM3399;
XX
DT 03-JUN-2004 (first entry)
XX
DE Human SCN5A variant 3 protein SEQ ID NO:6.
XX
KW human; cardiac sodium channel alpha subunit; SCN5A;
KW sodium channel-gated channel; type V; alpha; mutation study;
KW sodium channel related disease.
XX
OS Homo sapiens.
XX
PN MO2004012668-A2.
PD 12-FEB-2004.
XX
PF 01-AUG-2003; 2003WO-US024190.
XX
PR 02-AUG-2002; 2002US-0401018P.
XX
PA (WISC ) WISCONSIN ALUMNI RES FOUND.
PI Makieleki JC, Ye B, Ackerman MJ;
XX
DR WPI: 2004-157000/15.
XX
DR N-PEDB; ADM3398.
XX
PT New nucleic acid encoding sodium voltage-gated channel, type V, alpha
PT polypeptide variants, useful for studying mutations, and designing or
PT identifying new diagnostics and treatment strategies or agents for sodium
PT channel related diseases.
XX
PS Claim 14, SEQ ID NO 6, 11pp; English.
XX

The present sequence represents a variant of the human cardiac sodium
channel alpha subunit designated SCN5A (sodium voltage-gated channel,
type V, alpha) protein. The present invention describes an isolated
polynucleotide encoding an SCN5A polypeptide where the polynucleotide is
selected from the group: (1) a first polynucleotide that encodes an SCN5A
polypeptide selected from the group consisting of: (i) a histidine,
CC threonine, leucine, arginine and glutamine at amino acid positions 558,
CC 559, 618, 1027 and 1077, respectively; (ii) an arginine, threonine,
CC leucine, arginine and glutamine at amino acid positions 558, 559, 618,
CC 1027 and 1077, respectively; (iii) a histidine, threonine, leucine and
CC arginine at amino acid positions 558, 559, 618 and 1027, respectively,
CC with the amino acid at amino acid position 1077 deleted; or (iv) an
CC arginine, threonine, leucine and arginine at amino acid positions 558,
CC 559, 618 and 1027, respectively, with the amino acid at amino acid
CC position 1077 deleted; (2) a second polynucleotide that is at least 80 %
CC identical to the first polynucleotide over the entire length of the first
CC polypeptide; (3) a third polynucleotide that encodes any of the SCN5A
CC polypeptides with a conservative substitution, deletion or rearrangement
CC at one or more non-critical amino acid position; and (4) a fourth
CC polynucleotide that is a complement of the first, second or third
CC polynucleotide. Also described: (1) a genetic construct comprising the
CC polynucleotide operably linked to a non-native expression control
CC sequence; (2) a cell comprising the polynucleotide; (3) an isolated
CC polypeptide encoded by the polynucleotide; (4) an antibody that
CC specifically binds to the polypeptide; (5) identifying an agent that can
CC alter the activity of a sodium channel; (6) identifying an agent that can
CC alter the expression of a sodium channel; (7) determining whether a

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biological sample or a preparation derived from the biological sample contains the polypeptide; (8) determining whether a mutation on a sodium channel is associated with a disease; and (9) determining whether a human or non-human subject is at risk for long QT syndrome. The SCNSA polynucleotides and polypeptides are useful for studying mutations, and designing or identifying new diagnostics and treatment strategies or agents for sodium channel related diseases or conditions.

Sequence 2015 AA:

Query Match 99.9%; Score 10484; DB 8; Length 2015;
Best Local Similarity 99.9%; Pred. No. 0;
Matches 2014; Conservative 0; Mismatches 1; Indels 0; Gaps 0;

QY 1 MANFLPRGTSFRRTFRESLAIEKRMAEKQARGSTTLQESRGLPEEAPRPOLDIOA 60
DB 1 MANFLPRGTSFRRTFRESLAIEKRMAEKQARGSTTLQESRGLPEEAPRPOLDIOA 60
QY 61 SKKLPDLVGNPQELIGPELDLPDFYSTOKTFTLVNKGKTIIPRSATNALVYLSPPPI 120
DB 61 SKKLPDLVGNPQELIGPELDLPDFYSTOKTFTLVNKGKTIIPRSATNALVYLSPPPI 120
QY 121 RRAVKILVHSLFNNLIMCTILTNCFVMAQHPPEWTKYVEYTFATVTFESLVKILARG 180
DB 121 RRAVKILVHSLFNNLIMCTILTNCFVMAQHPPEWTKYVEYTFATVTFESLVKILARG 180
QY 121 RRAVKILVHSLFNNLIMCTILTNCFVMAQHPPEWTKYVEYTFATVTFESLVKILARG 180
DB 121 RRAVKILVHSLFNNLIMCTILTNCFVMAQHPPEWTKYVEYTFATVTFESLVKILARG 180
QY 181 FCLHAFTELDPWNNLDFSVIIMAYTTEFVDLGNVSALRTRVRLPAKTTISVIGLKTIV 240
DB 181 FCLHAFTELDPWNNLDFSVIIMAYTTEFVDLGNVSALRTRVRLPAKTTISVIGLKTIV 240
QY 241 GALLQSVKRLADVWMLTVFCISVFPALIGLOLFMGMLRHKCVRNFPALNGTNGSEADGLV 300
DB 241 GALLQSVKRLADVWMLTVFCISVFPALIGLOLFMGMLRHKCVRNFPALNGTNGSEADGLV 300
QY 301 WESLDLYSDPENNYLLKNGTSDVLLCGNSSDAGTCEGYRCLKAGENDHGYTSFDSFAM 360
DB 301 WESLDLYSDPENNYLLKNGTSDVLLCGNSSDAGTCEGYRCLKAGENDHGYTSFDSFAM 360
QY 361 AFLALFRLMTODCWERLYOQTLRSAGKIYMFMLVIFLGSFYLVNLLAVANAYEON 420
DB 361 AFLALFRLMTODCWERLYOQTLRSAGKIYMFMLVIFLGSFYLVNLLAVANAYEON 420
QY 421 QATTAETBEKEKRFQEAEMMLKKEHEALTIRGVTVSSLSMSPLAVNSHERSKRK 480
DB 421 QATTAETBEKEKRFQEAEMMLKKEHEALTIRGVTVSSLSMSPLAVNSHERSKRK 480
QY 481 RMSSGTECGEDRLPKSDESDGPRAMNHLSTRGLSRTSMKPRSSRGSIIFTRRRDLGSE 540
DB 481 RMSSGTECGEDRLPKSDESDGPRAMNHLSTRGLSRTSMKPRSSRGSIIFTRRRDLGSE 540
QY 541 ADPADENSTAGESSESHTSLVPMPLRRTSAQOGPSPTSAFGHALHGKKNSTYDCGV 600
DB 541 ADPADENSTAGESSESHTSLVPMPLRRTSAQOGPSPTSAFGHALHGKKNSTYDCGV 600
QY 601 VSLDAGDPEATSPGSHLRPWLHPPTTTPSEPGGPOMLTJQAPCVDSFEPPGAKQ 660
DB 601 VSLDAGDPEATSPGSHLRPWLHPPTTTPSEPGGPOMLTJQAPCVDSFEPPGAKQ 660
QY 661 RALSASVLTSLAELEESRHKPCPCMNLAQRYLIWECCPLAMSIKOGVKLVMDPFTD 720
DB 661 RALSASVLTSLAELEESRHKPCPCMNLAQRYLIWECCPLAMSIKOGVKLVMDPFTD 720
QY 721 LITTCIVLNTLPMALSHYNTSEPEEMLOVGNLVFTGIFTAEMTKIITADPYYPQOG 780
DB 721 LITTCIVLNTLPMALSHYNTSEPEEMLOVGNLVFTGIFTAEMTKIITADPYYPQOG 780
QY 781 WNIIPSIIVTSLMEJGSRMSNLSVLRFRLLRYPKLAKSWPTNTLTKIIGNSVGALG 840
DB 781 WNIIPSIIVTSLMEJGSRMSNLSVLRFRLLRYPKLAKSWPTNTLTKIIGNSVGALG 840
QY 841 NTLVLAIVTFAVVGMQLFGKNYSELSDSGLLPRWHMDFPHAFLIFRIICGEVI 900
DB 841 NTLVLAIVTFAVVGMQLFGKNYSELSDSGLLPRWHMDFPHAFLIFRIICGEVI 900

QY 901 ETMMDCHEVSGOSTCLLVFLVWVIGNLVYVNLFLALILSSFSADNLTAPEDEKENNLQ 960
DB 901 ETMMDCHEVSGOSTCLLVFLVWVIGNLVYVNLFLALILSSFSADNLTAPEDEKENNLQ 960
QY 961 LALARIORGRLFYKRTTMDPCCGLLRQRPQKAPALAAQQLPSCIAITPYSPPPETEKVP 1020
DB 961 LALARIORGRLFYKRTTMDPCCGLLRQRPQKAPALAAQQLPSCIAITPYSPPPETEKVP 1020
QY 1021 PTRKETRFEEGEPGCGTPEDEPVCYPIAIVASDTDDQEDENSLGTEESKQESOP 1080
DB 1021 PTRKETRFEEGEPGCGTPEDEPVCYPIAIVASDTDDQEDENSLGTEESKQESOP 1080
QY 1081 VSGPEAPDPDSRTMSQVATASSEAEASASQAMRQOMKAEPOAPGCGTEPESSCEBGT 1140
DB 1081 VSGPEAPDPDSRTMSQVATASSEAEASASQAMRQOMKAEPOAPGCGTEPESSCEBGT 1140
QY 1141 ADMNTTAELEQIPDLGQDYKDEDCFTTEGCVRRCPCAVDTTQAPGKVMRLKTCYHI 1200
DB 1141 ADMNTTAELEQIPDLGQDYKDEDCFTTEGCVRRCPCAVDTTQAPGKVMRLKTCYHI 1200
QY 1201 VEHSMFTFTIIFMILSSGALAEEDYLERKRTIKVLEBYADKMTFTYVFLMELKWAY 1260
DB 1201 VEHSMFTFTIIFMILSSGALAEEDYLERKRTIKVLEBYADKMTFTYVFLMELKWAY 1260
QY 1261 GFKKYFTNACWMLDPLVDVSLVSVANTLGFPAEMGPKLSRTLRLALRPLALSREGMR 1320
DB 1261 GFKKYFTNACWMLDPLVDVSLVSVANTLGFPAEMGPKLSRTLRLALRPLALSREGMR 1320
QY 1321 VVNVNLAVALPSIMNVLLVCLIFMLIFSINGVNLPAKFGRCINQTEGDIPLNTTYVNNK 1380
DB 1321 VVNVNLAVALPSIMNVLLVCLIFMLIFSINGVNLPAKFGRCINQTEGDIPLNTTYVNNK 1380
QY 1381 SOCESLNLTEGLYTKKRVNPDVNGAGYLLAQVATFKGMDIMYAVDSRGYEOQWE 1440
DB 1381 SOCESLNLTEGLYTKKRVNPDVNGAGYLLAQVATFKGMDIMYAVDSRGYEOQWE 1440
QY 1441 YNTLYMYTIVPIIFIGSFFTLNLFIGIYIDNPNQKKLGGODIEMTEBQKXTYNNAMKUL 1500
DB 1441 YNTLYMYTIVPIIFIGSFFTLNLFIGIYIDNPNQKKLGGODIEMTEBQKXTYNNAMKUL 1500
QY 1501 GSKKQKPIPRPLNKYOGFIIDYTKQAFDVTIMFLCLNMVTMMVETDDQSEPKINILA 1560
DB 1501 GSKKQKPIPRPLNKYOGFIIDYTKQAFDVTIMFLCLNMVTMMVETDDQSEPKINILA 1560
QY 1561 KINLLFVAITGECIVKLAALRHYYFPNNSWNIIDPVVVILISTGYTUSDIIQKTFSPTL 1620
DB 1561 KINLLFVAITGECIVKLAALRHYYFPNNSWNIIDPVVVILISTGYTUSDIIQKTFSPTL 1620
QY 1621 FFRVRLARIGIRLIRGAGIRTLPALMMSLPALFNIGLLFLVWFYISFGMANFAY 1680
DB 1621 FFRVRLARIGIRLIRGAGIRTLPALMMSLPALFNIGLLFLVWFYISFGMANFAY 1680
QY 1681 VKMEAGIDDMFNQFTFANSMCLFOITTSAGMDGLSPIINTGPYCDPTLPNSNGSRGD 1740
DB 1681 VKMEAGIDDMFNQFTFANSMCLFOITTSAGMDGLSPIINTGPYCDPTLPNSNGSRGD 1740
QY 1741 CGSPAVGILFFTTYIIISFLVNMVTAIILENSVATTESTEPLSDDDDMFEIWEKF 1800
DB 1741 CGSPAVGILFFTTYIIISFLVNMVTAIILENSVATTESTEPLSDDDDMFEIWEKF 1800
QY 1801 DPEATOFIEVSVDSPADALSEPLRIAKPNQISLIINDLPMVSGDRHCHMDILFAFTKRV 1860
DB 1801 DPEATOFIEVSVDSPADALSEPLRIAKPNQISLIINDLPMVSGDRHCHMDILFAFTKRV 1860
QY 1861 LGESGENDALKIQHEEKFMANPSKISYEPITTTLRKHEEVSAMVIQAPRRHLQRL 1920
DB 1861 LGESGENDALKIQHEEKFMANPSKISYEPITTTLRKHEEVSAMVIQAPRRHLQRL 1920
QY 1921 KHASFLFRQOAGSGISEBDAERGLIAYVMSFNRPPLGPPSSSISSTSPSSYSVT 1980
DB 1921 KHASFLFRQOAGSGISEBDAERGLIAYVMSFNRPPLGPPSSSISSTSPSSYSVT 1980

QY 1981 RATSNDLQVSGSDYSHSEDLADFPSPDRRESIV 2015
 DB 1981 RATSNDLQVSGSDYSHSEDLADFPSPDRRESIV 2015

RESULT 4
 AEB22972
 ID AEB22972 standard; protein; 2015 AA.
 XX AEB22972;
 AC
 XX
 DT 22-SEP-2005 (fixed entry)
 XX
 DE Sodium channel alpha subunit human SCN5A gene derived protein.
 XX
 KW SCN5A; sodium channel; Brugada syndrome; heart arrhythmia;
 XX antiarrhythmic; selectable marker; ventricular fibrillation.
 OS Homo sapiens.
 XX
 PN JP2005192413-A.
 XX
 PD 21-JUL-2005.
 XX
 PF 26-DEC-2003; 2003JP-00435234.
 XX
 PR 26-DEC-2003; 2003JP-00435234.
 XX
 PA (DOKU-) DOKURITSU GYOSEI HOJIN KAGAKU GIJUTSU SH.
 XX
 PI Matenaga A;
 XX
 PI WPI; 2005-501993/51.
 DR
 XX
 PT Novel SCN5A gene of sodium channel alpha subunit, with mutations in which
 PT glycine being substituted by serine, and serine being substituted by
 PT leucine, at specific positions, useful as marker for diagnosing Brugada
 PT syndrome.
 XX
 PS Disclosure; SEQ ID NO 1; 30pp; Japanese.
 XX
 CC The invention relates to a novel SCN5A gene of a sodium channel alpha
 CC subunit. The novel SCN5A gene comprises the mutation G292S between the
 CC fifth and sixth membrane passing-through subunits of the first domain,
 CC and/or the mutation S835L of the intracellular loop between the fourth
 CC and fifth membrane passing-through subunits of the second domain. The
 CC novel SCN5A gene can be used as a Brugada syndrome marker, for diagnosing
 CC Brugada syndrome. Brugada syndrome causes idiopathic ventricular
 CC fibrillation, thus the gene/marker allow for the prevention or treatment
 CC of the syndrome. This sequence represents the protein of the wild-type
 CC human SCN5A gene of the invention.
 CC
 XX
 SQ Sequence 2015 AA;

Query Match 99.9%; Score 10484; DB 9; Length 2015;
 Best Local Similarity 99.9%; Pred. No. 0; Mismatches 1; Indels 0; Gaps 0;
 Match 2014; Conservative 0; Mismatches 1; Indels 0; Gaps 0;

QY 1 MANFLPRTGTSFRFTRESLALAEKMAEKQARGSTTLQESREGLPPEEAPRPOLDIOA 60
 DB 1 MANFLPRTGTSFRFTRESLALAEKMAEKQARGSTTLQESREGLPPEEAPRPOLDIOA 60
 QY 61 SKGLPDLGNPPQELIGEPLEDDLPFYSTOKTFYLANAKTIFRPSATNALVYLSPPHPI 120
 DB 61 SKGLPDLGNPPQELIGEPLEDDLPFYSTOKTFYLANAKTIFRPSATNALVYLSPPHPI 120
 QY 121 RRAAVKIIVHSLFNNLIMCTITLNCVFMAOHDPPTWTKYVEYTFATITPESLVYILARG 180
 DB 121 RRAAVKIIVHSLFNNLIMCTITLNCVFMAOHDPPTWTKYVEYTFATITPESLVYILARG 180
 QY 181 FCLHAFTELDPWMNLDSFVIIIMAYTTEFVDLGNVSALRTPFVLAALXTISVIGLAKTIV 240
 DB 181 FCLHAFTELDPWMNLDSFVIIIMAYTTEFVDLGNVSALRTPFVLAALXTISVIGLAKTIV 240

QY 241 GALIOSYKKADAVWVLTJYFCLSVFALIGLQFMGNLRHKCYANFTALANGTNGSV EADGLV 300
 DB 241 GALIOSYKKADAVWVLTJYFCLSVFALIGLQFMGNLRHKCYANFTALANGTNGSV EADGLV 300
 QY 301 WESLDLYLSDPENYLLKNGTSDVLLCGNSSDAGTCPEGYRCUKAGBNPDHGYTSFDSFAM 360
 DB 301 WESLDLYLSDPENYLLKNGTSDVLLCGNSSDAGTCPEGYRCUKAGBNPDHGYTSFDSFAM 360
 QY 361 AFLALFRLMTQDCWERYLQOTLRAGKTYMIFMLVILGSPYLVNLIAYVAMAYEON 420
 DB 361 AFLALFRLMTQDCWERYLQOTLRAGKTYMIFMLVILGSPYLVNLIAYVAMAYEON 420
 QY 421 QATIAETEKEKRPCEANEMMLKKEHEALTIRGVTVSSLSLEMSPLAPNHSERSKRX 480
 DB 421 QATIAETEKEKRPCEANEMMLKKEHEALTIRGVTVSSLSLEMSPLAPNHSERSKRX 480
 QY 481 RMSGTEECGEDRLPKSDSEDPGRAMNHLSTRGLSRTSMKRSRSGSIFTRRRDLGSE 540
 DB 481 RMSGTEECGEDRLPKSDSEDPGRAMNHLSTRGLSRTSMKRSRSGSIFTRRRDLGSE 540
 QY 541 ADPADDENSTAGESESHRTSLVWPILRRTSAQCQSPGTSAPGHALHGXNSTDVDCNV 600
 DB 541 ADPADDENSTAGESESHRTSLVWPILRRTSAQCQSPGTSAPGHALHGXNSTDVDCNV 600
 QY 601 VSLGAGPPEATSPGSHLLRPVMLEHPDPTTPSSEBPGPOMLTQACVDFEERGARQ 660
 DB 601 VSLGAGPPEATSPGSHLLRPVMLEHPDPTTPSSEBPGPOMLTQACVDFEERGARQ 660
 QY 661 RALSASVLTALAELESRRKCPQCNRLAQRVLIWECCLPMNSIKQGVKLVMDPFTD 720
 DB 661 RALSASVLTALAELESRRKCPQCNRLAQRVLIWECCLPMNSIKQGVKLVMDPFTD 720
 QY 721 LTTWCIYVANTLFMALHYNNTSEFEEMLOQGNLVFTGI FTAEWTFKIIALDPYYTFOG 780
 DB 721 LTTWCIYVANTLFMALHYNNTSEFEEMLOQGNLVFTGI FTAEWTFKIIALDPYYTFOG 780
 QY 781 WNIPISTIIVIIISLWELGSRMSNLSVLRSPFLRLRPFLAKSMPTNTLIIKIIGNSVGALG 840
 DB 781 WNIPISTIIVIIISLWELGSRMSNLSVLRSPFLRLRPFLAKSMPTNTLIIKIIGNSVGALG 840
 QY 841 NLTVLAIIVFIFAVVGQVLFQKNYSLELRDSDGLLPRMHMDFFHAFLIFRILCGEMI 900
 DB 841 NLTVLAIIVFIFAVVGQVLFQKNYSLELRDSDGLLPRMHMDFFHAFLIFRILCGEMI 900
 QY 901 ETMMDCEVSGQSCLVFLVWYIGNLVINLFLALILSFRSADNLTAPEDREMNLIQ 960
 DB 901 ETMMDCEVSGQSCLVFLVWYIGNLVINLFLALILSFRSADNLTAPEDREMNLIQ 960
 QY 961 LALARIQGLRFVYKRTTWDGCCGLLRORPQRPALAAAGOLPSCITATYSPPPETEXVP 1020
 DB 961 LALARIQGLRFVYKRTTWDGCCGLLRORPQRPALAAAGOLPSCITATYSPPPETEXVP 1020
 QY 1021 PTRKRETBEGEOPGQGTGPDPEPCVPIAAESPTDQOEDEBENSIGTEBESSSQOESOP 1080
 DB 1021 PTRKRETBEGEOPGQGTGPDPEPCVPIAAESPTDQOEDEBENSIGTEBESSSQOESOP 1080
 QY 1081 VSGGPBAPPDSTRTSQVSATASSEAEASASQADWRQKAPQAPGCGETEDSCSEST 1140
 DB 1081 VSGGPBAPPDSTRTSQVSATASSEAEASASQADWRQKAPQAPGCGETEDSCSEST 1140
 QY 1141 ADMNTNLAELQIPDLGQVDPEDCFTEGCVRRCPCCAVDTTQAPGVWRRLRKTCHYI 1200
 DB 1141 ADMNTNLAELQIPDLGQVDPEDCFTEGCVRRCPCCAVDTTQAPGVWRRLRKTCHYI 1200
 QY 1201 VEHSMFETFTIFMLILSSGALAFEDYIEBKRTIKVLEIYADKQFTYFVLEMLIKWAY 1260
 DB 1201 VEHSMFETFTIFMLILSSGALAFEDYIEBKRTIKVLEIYADKQFTYFVLEMLIKWAY 1260
 QY 1261 GFKKYFTNAMCMLDPLIVDSVSLVANTLGFAEMGPIKSLRTLRALRPLALSREFGMR 1320
 DB 1261 GFKKYFTNAMCMLDPLIVDSVSLVANTLGFAEMGPIKSLRTLRALRPLALSREFGMR 1320

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QY 1321 VVNAALGATPSIMNVLVCLIFWLIISIMGVNLPAKFGRCINOTGEDLPLANTIVNNK 1380
DB 1321 VVNAALGATPSIMNVLVCLIFWLIISIMGVNLPAKFGRCINOTGEDLPLANTIVNNK 1380
QY 1381 SCSLSNLTGELVYTKVNFNDNGAGYATALLQVATFKNMDIMVAAVDSRGVEQOPWE 1440
DB 1381 SCSLSNLTGELVYTKVNFNDNGAGYATALLQVATFKNMDIMVAAVDSRGVEQOPWE 1440
QY 1441 YNLVYVIFVIFIIIFGSEFTNLPIGVIINDPNQKKLGGODIPMTBEQKKYNNAKKL 1500
DB 1441 YNLVYVIFVIFIIIFGSEFTNLPIGVIINDPNQKKLGGODIPMTBEQKKYNNAKKL 1500
QY 1501 GSKXPQKPIPRPLNKYQGFIDIVTKQAFDVTIMFLICLNAVTVMMVEEDDQSPKINILA 1560
DB 1501 GSKXPQKPIPRPLNKYQGFIDIVTKQAFDVTIMFLICLNAVTVMMVEEDDQSPKINILA 1560
QY 1561 KINILFVAIFGCECIVKLAALRHYYFTNSMNI FDEVVILSI VGTVLSDIIQKTFPSPTL 1620
DB 1561 KINILFVAIFGCECIVKLAALRHYYFTNSMNI FDEVVILSI VGTVLSDIIQKTFPSPTL 1620
QY 1621 FRVIRLARIIGRLIRIRGAKGIRTLFPALMMSLPALFNIGLLFLVWFYISIFGMANFAY 1680
DB 1621 FRVIRLARIIGRLIRIRGAKGIRTLFPALMMSLPALFNIGLLFLVWFYISIFGMANFAY 1680
QY 1681 VKWEGCIDDMENPQTFANSMCLFOITTSAGMDGLSPILMTGPPYCDPTLPNSGSRGD 1740
DB 1681 VKWEGCIDDMENPQTFANSMCLFOITTSAGMDGLSPILMTGPPYCDPTLPNSGSRGD 1740
QY 1741 CGSPAVGILFTTYYIIISFLIVNNVYIAIILENFSVATEESTEPLESDPFMFYEIWEKF 1800
DB 1741 CGSPAVGILFTTYYIIISFLIVNNVYIAIILENFSVATEESTEPLESDPFMFYEIWEKF 1800
QY 1801 DPEATQFIEYSVLSDFADALSEPLRIAKPNQISLINMDLPVNSGRHICMDILPAFTKRV 1860
DB 1801 DPEATQFIEYSVLSDFADALSEPLRIAKPNQISLINMDLPVNSGRHICMDILPAFTKRV 1860
QY 1861 LGSESEMALKIOMEKEKPMANPSKISYEPIITTLRRKHEEVSAMVIOARFRHLLQNSL 1920
DB 1861 LGSESEMALKIOMEKEKPMANPSKISYEPIITTLRRKHEEVSAMVIOARFRHLLQNSL 1920
QY 1921 KHASELFRQOAGSGISEDAPEREGLIIVVSENFSPRLGPPSSSSISSTSPPSYDSVT 1980
DB 1921 KHASELFRQOAGSGISEDAPEREGLIIVVSENFSPRLGPPSSSSISSTSPPSYDSVT 1980
QY 1981 RATSNDLQVRGSDYSHSEDLADFPSPSPDRRESIV 2015
DB 1981 RATSNDLQVRGSDYSHSEDLADFPSPSPDRRESIV 2015

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RESULT 5

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ADM33997
ID ADM33997 standard; protein; 2016 AA.
AC ADM33997;
DT 03-JUN-2004 (first entry)
DE Human SCN5A variant 2 protein SEQ ID NO:4.
XX human; cardiac sodium channel; alpha subunit; SCN5A;
XX sodium voltage-gated channel; type V; alpha; mutation study;
XX sodium channel related disease.
XX Homo sapiens.
XX MO2004012668-A2.
XX 12-FEB-2004.
XX 01-AUG-2003; 2003WO-US024190.
XX 02-AUG-2002; 2002US-0401018P.
XX

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PA (MISC ) WISCONSIN ALUMNI RES FOUND.
XX
PI Makielaki UC, Ye B, Ackerman MJ;
XX
DR WPI; 2004-157000/15.
XX N-PSDB; ADM33996.
XX
PT New nucleic acid encoding sodium voltage-gated channel, type V, alpha
PT polypeptide variants, useful for studying mutations, and designing or
PT identifying new diagnostics and treatment strategies or agents for sodium
PT channel related diseases.
XX
PS Claim 11; SEQ ID NO 4; 111pp; English.
XX
XX The present sequence represents a variant of the human cardiac sodium
CC channel alpha subunit designated SCN5A (sodium voltage-gated channel,
CC type V, alpha) protein. The present invention describes an isolated
CC polynucleotide encoding an SCN5A polypeptide where the polynucleotide is
CC selected from the group: (1) a first polynucleotide that encodes an SCN5A
CC polypeptide selected from the group consisting of: (i) a histidine,
CC threonine, leucine, arginine and glutamine at amino acid positions 558,
CC 559, 618, 1027 and 1077, respectively; (ii) an arginine, threonine,
CC leucine, arginine and glutamine at amino acid positions 558, 559, 618,
CC 1027 and 1077, respectively; (iii) a histidine, threonine, leucine and
CC arginine at amino acid positions 558, 559, 618 and 1027, respectively,
CC with the amino acid at amino acid position 1077 deleted; or (iv) an
CC arginine, threonine, leucine and arginine at amino acid positions 558,
CC 559, 618 and 1027, respectively, with the amino acid at amino acid
CC position 1077 deleted; (2) a second polynucleotide that is at least 80 %
CC identical to the first polynucleotide over the entire length of the first
CC polypeptide; (3) a third polynucleotide that encodes any of the SCN5A
CC polypeptides with a conservative substitution, deletion or rearrangement
CC at one or more non-critical amino acid position; and (4) a fourth
CC polynucleotide that is a complement of the first, second or third
CC polynucleotide. Also described: (1) a genetic construct comprising the
CC polynucleotide operably linked to a non-native expression control
CC sequence; (2) a cell comprising the polynucleotide; (3) an isolated
CC polypeptide encoded by the polynucleotide; (4) an antibody that
CC specifically binds to the polypeptide; (5) identifying an agent that can
CC alter the activity of a sodium channel; (6) identifying an agent that can
CC alter the expression of a sodium channel; (7) determining whether a
CC biological sample or a preparation derived from the biological sample
CC contains the polypeptide; (8) determining whether a mutation on a sodium
CC channel is associated with a disease; and (9) determining whether a human
CC or non-human subject is at risk for Long QT syndrome. The SCN5A
CC polynucleotides and polypeptides are useful for studying mutations, and
CC designing or identifying new diagnostics and treatment strategies or
CC agents for sodium channel related diseases or conditions.
XX
SQ Sequence 2016 AA;

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Query Match 99.9%; Score 10478.5; DB 8; Length 2016;
Best Local Similarity 99.9%; Pred. No. 0; Mismatches 0; Indels 1; Gaps 1;
Matches 2015; Conservative 0;

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QY 1 MANFLPBGTSPPRFTRESLAATEKMAEKQAGSTTIOBSRGLEPEEBAPRQDLOA 60
DB 1 MANFLPBGTSPPRFTRESLAATEKMAEKQAGSTTIOBSRGLEPEEBAPRQDLOA 60
QY 61 SKKLPDIYGNPQELIGEPLEDDPFYSTQKTFIVLNKKTIFFFSATNALYVSPHPPI 120
DB 61 SKKLPDIYGNPQELIGEPLEDDPFYSTQKTFIVLNKKTIFFFSATNALYVSPHPPI 120
QY 121 RRAAVKIIVHSLFNNLIMCTILTNVCVMAQOHDPDPMPKRYEYVETATAYTFESIVKILARG 180
DB 121 RRAAVKIIVHSLFNNLIMCTILTNVCVMAQOHDPDPMPKRYEYVETATAYTFESIVKILARG 180
QY 181 FCLHAFPLRDPKMMLDPSVITIMAYTFEFVDLGNVSLARTFRVLRALKTISVIGLKTIV 240
DB 181 FCLHAFPLRDPKMMLDPSVITIMAYTFEFVDLGNVSLARTFRVLRALKTISVIGLKTIV 240
QY 241 TALLIOSVKKGLADVWVLTVFCLSVPALIGLQLEFNGNLRHRCVARNFTALNGTNGSYEADGLV 300

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Db 241 GALLIOSVKKLADVWVLTVECLSVFALIGLQFMGNLRHKCNFTALNTNGSVSEADGLV 300
 QY WESLXYLSDPENNYLTKNTSDVLLCGNSSDAGTCEGRCLKAGNPBGHTSPSPFW 360
 Db 301 WESLXYLSDPENNYLTKNTSDVLLCGNSSDAGTCEGRCLKAGNPBGHTSPSPFW 360
 QY 361 AFLALFRLMTQOCMERLYOQTLRSAGKIYIMIFMLYIFLGSFYLVLILAVAMAAYEON 420
 Db 361 AFLALFRLMTQOCMERLYOQTLRSAGKIYIMIFMLYIFLGSFYLVLILAVAMAAYEON 420
 QY 421 QATIAETEBEKRRFOEAMEMLKKEHEALTIRGVDIVSRSSLEMSPLAPVNSHERSKRRK 480
 Db 421 QATIAETEBEKRRFOEAMEMLKKEHEALTIRGVDIVSRSSLEMSPLAPVNSHERSKRRK 480
 QY 481 RMSSGTEECGECRLPSPSDESDGPRANNHLSLRGSLSRSMKPRSSSGSIPTRRRDLSGE 540
 Db 481 RMSSGTEECGECRLPSPSDESDGPRANNHLSLRGSLSRSMKPRSSSGSIPTRRRDLSGE 540
 QY 541 ADPADENSTAGESESHRTSLVPMPLRRTSAQOQSPGTSAPGHALHKKNSYVDCNGV 600
 Db 541 ADPADENSTAGESESHRTSLVPMPLRRTSAQOQSPGTSAPGHALHKKNSYVDCNGV 600
 QY 601 VSLGAGDEBEATSPGSHLLRPVMLBHPDPTTTPSEBPGGPOMLTSQAPCVDGFEEBGAQ 660
 Db 601 VSLGAGDEBEATSPGSHLLRPVMLBHPDPTTTPSEBPGGPOMLTSQAPCVDGFEEBGAQ 660
 QY 661 RALSAVSULTSLBEBESRHKCPRCMRLAORYLIWECCPLMMSIKQVYKLVNDPFD 720
 Db 661 RALSAVSULTSLBEBESRHKCPRCMRLAORYLIWECCPLMMSIKQVYKLVNDPFD 720
 QY 721 LTTMCIYVANTLPMALBHNMTSBPEBMQVGNLFTGIFTAEMTKIALDPYYPFOG 780
 Db 721 LTTMCIYVANTLPMALBHNMTSBPEBMQVGNLFTGIFTAEMTKIALDPYYPFOG 780
 QY 781 WNIPIISIIYLSLMEIGLSRMSNLSTYLSFRLRYFKLAKSWPTLNTLIIKIGNSYGALG 840
 Db 781 WNIPIISIIYLSLMEIGLSRMSNLSTYLSFRLRYFKLAKSWPTLNTLIIKIGNSYGALG 840
 QY 841 NITLVLAIVTFPANYGMQLFGKNTSELDSGSLPRMNMDFHAFILITRIICGEVI 900
 Db 841 NITLVLAIVTFPANYGMQLFGKNTSELDSGSLPRMNMDFHAFILITRIICGEVI 900
 QY 901 ETTMDCMEVSGOSCLVFLVLMVIGNLVNLFLALILSSPSADNLTPADEDRMNNQ 960
 Db 901 ETTMDCMEVSGOSCLVFLVLMVIGNLVNLFLALILSSPSADNLTPADEDRMNNQ 960
 QY 961 LALARIORGRLRFVKRTTMDFCGGLRORPOKPAALAAQOLPSCIATPYSPPEPEKVP 1020
 Db 961 LALARIORGRLRFVKRTTMDFCGGLRORPOKPAALAAQOLPSCIATPYSPPEPEKVP 1020
 QY 1021 PTRKETREBEQPOQGTIPGDEPVCPIAVAESDTDOEEDENS LGTEBESSK-QESQ 1079
 Db 1021 PTRKETREBEQPOQGTIPGDEPVCPIAVAESDTDOEEDENS LGTEBESSK-QESQ 1080
 QY 1080 PVSQGPAPPDSTRITMSOVSATSSSEAEASASQADRMQOKAPQAPGCGETEDSCSBS 1139
 Db 1081 PVSQGPAPPDSTRITMSOVSATSSSEAEASASQADRMQOKAPQAPGCGETEDSCSBS 1140
 QY 1140 TADMTNTAELLEQIPDLGQVDVPEDCEFTGECVRRCPCCAVDTTQAPGVWMLRKTCTYH 1199
 Db 1141 TADMTNTAELLEQIPDLGQVDVPEDCEFTGECVRRCPCCAVDTTQAPGVWMLRKTCTYH 1200
 QY 1200 IVEHSMFETFIIFMILLSSGALAFBDIYIEBKTKITVLEVDKMTTYFVLEMLIKMYA 1259
 Db 1201 IVEHSMFETFIIFMILLSSGALAFBDIYIEBKTKITVLEVDKMTTYFVLEMLIKMYA 1260
 QY 1260 YGFKKYFTNAMCMLDFLIVDSVLSVANTLGFAMGPKISLRTLRALRPLALSRPESM 1319
 Db 1261 YGFKKYFTNAMCMLDFLIVDSVLSVANTLGFAMGPKISLRTLRALRPLALSRPESM 1320
 QY 1320 RYVVALVGAISINMVLVLCILFMLIFSIMGVNLFAKFGRCINOTEGDPLANTYVNN 1379
 Db 1321 RYVVALVGAISINMVLVLCILFMLIFSIMGVNLFAKFGRCINOTEGDPLANTYVNN 1380

QY 1380 KSQCESLNLITGELTYMTKVKVNFNDVGAAGYIALLOVATFGKMDIMYAADVSRGYEOPOM 1439
 Db 1381 KSQCESLNLITGELTYMTKVKVNFNDVGAAGYIALLOVATFGKMDIMYAADVSRGYEOPOM 1440
 QY 1440 EYNLYMYIYFVFIIFISGSEFTLNLFIYIINDFNQOKKKGGQDIFMTEBOKKYYNAMK 1499
 Db 1441 EYNLYMYIYFVFIIFISGSEFTLNLFIYIINDFNQOKKKGGQDIFMTEBOKKYYNAMK 1500
 QY 1500 LGSKKPQKPIPRPLANKYGFIFDIYTKQAFDVTIMFLICLANNVTMVEITDDSPKINIL 1559
 Db 1501 LGSKKPQKPIPRPLANKYGFIFDIYTKQAFDVTIMFLICLANNVTMVEITDDSPKINIL 1560
 QY 1560 AKINLFAVIFGECIVKLAALRHYFPNSMNIIPFVYVILISVGTVLSDIIOKTFEFT 1619
 Db 1561 AKINLFAVIFGECIVKLAALRHYFPNSMNIIPFVYVILISVGTVLSDIIOKTFEFT 1620
 QY 1620 LFRVIRLARIGRIILIRGAKGIRTLPLALMMSLPALFNIGLLFLVMEIYSIFGMANPA 1679
 Db 1621 LFRVIRLARIGRIILIRGAKGIRTLPLALMMSLPALFNIGLLFLVMEIYSIFGMANPA 1680
 QY 1680 YKMEAGIDDMNFOTFANSMLCLFOITTSAGMDGLSPILNTGPPYCDPTLPNSGSRG 1739
 Db 1681 YKMEAGIDDMNFOTFANSMLCLFOITTSAGMDGLSPILNTGPPYCDPTLPNSGSRG 1740
 QY 1740 DCGSPAVGILFETTYIIISPLIVNMVYAIILNENSVATESSTEBLSEDDPMFEIWEK 1799
 Db 1741 DCGSPAVGILFETTYIIISPLIVNMVYAIILNENSVATESSTEBLSEDDPMFEIWEK 1800
 QY 1800 FDPBEATQFIEYSVLSDFADALSEPRLAKPNQISLINDLPVSGDRHCHMDILFAFTGR 1859
 Db 1801 FDPBEATQFIEYSVLSDFADALSEPRLAKPNQISLINDLPVSGDRHCHMDILFAFTGR 1860
 QY 1860 VLGESGEMDALKIOMEKFPMAANPSKISYEPTITTLRRKHEEVSAMVIOARFRHLORS 1919
 Db 1861 VLGESGEMDALKIOMEKFPMAANPSKISYEPTITTLRRKHEEVSAMVIOARFRHLORS 1920
 QY 1920 LKHAFLFRQOAGSLSEEDAPEREGLIAYVMSNFSPPLGPPSSSISSTSPSPSYSV 1979
 Db 1921 LKHAFLFRQOAGSLSEEDAPEREGLIAYVMSNFSPPLGPPSSSISSTSPSPSYSV 1980
 QY 1980 TRATSDNLQVRGSDYSHSEDLADFPSPDRDRESIV 2015
 Db 1981 TRATSDNLQVRGSDYSHSEDLADFPSPDRDRESIV 2016

RESULT 6
 AEB22993
 ID AEB22993 standard; protein; 2015 AA.
 XX
 AC AEB22993;
 XX
 AC 22-SEP-2005 (first entry)
 DT
 XX
 DE Mutant human SCNSA protein, S835L.
 KW SCNSA; sodium channel; Brugada syndrome; heart arrhythmia;
 XX antiarrhythmic; selectable marker; ventricular fibrillation; mutein.
 OS Homo sapiens.
 OS Synthetic.
 XX
 FT Key Location/Qualifiers
 FT Misc-difference 835
 FT /note= "The wild-type Ser residue has been substituted
 FT with Leu"
 FT
 PN JP2005192413-A.
 XX
 PD 21-JUL-2005.
 XX
 PF 26-DEC-2003; 2003JP-00435234.
 XX

PR 26-DEC-2003; 2003JP-00435234.
XX
XX (DOKU-) DOKURITSU GYOSEI HOJIN KAGAKU GIJUTSU SH.
XX
XX Matsunaga A;
PI
PI WPI; 2005-501993/51.
DR
XX Novel SCN5A gene of sodium channel alpha subunit, with mutations in which
PT glycine being substituted by serine, and serine being substituted by
PT leucine, at specific positions, useful as marker for diagnosing Brugada
PT syndrome.
PS
XX Claim 1; Page: 30pp; Japanese.
XX
XX The invention relates to an novel SCN5A gene of a sodium channel alpha
CC subunit. The novel SCN5A gene comprises the mutation G292S between the
CC fifth and sixth membrane passing-through subunits of the first domain;
CC and/or the mutation S835L of the intracellular loop between the fourth
CC and fifth membrane passing-through subunits of the second domain. The
CC novel SCN5A gene can be used as a Brugada syndrome marker, for diagnosing
CC Brugada syndrome. Brugada syndrome causes idiopathic ventricular
CC fibrillation, thus the gene/marker allow for the prevention or treatment
CC of the syndrome. This sequence represents the S835L mutant protein of the
CC human SCN5A gene of the invention.
XX
SQ Sequence 2015 AA;
Query Match 99.9%; Score 10478; DB 9; Length 2015;
Best Local Similarity 99.9%; Pred. No. 0;
Matches 2013; Conservative 0; Mismatches 2; Indels 0; Gaps 0;
QY 1 MANFLPRTGSFRRFTBSLAIRKMAEKQARSTLQSRREGLPREEAPRPOLDIOA 60
DB 1 MANFLPRTGSFRRFTBSLAIRKMAEKQARSTLQSRREGLPREEAPRPOLDIOA 60
QY 61 SKKLVDLGNPQOELIGEPLEDLPFYSTOKTEIYLVNKGKTI FRFSATNALVYLSPPFI 120
DB 61 SKKLVDLGNPQOELIGEPLEDLPFYSTOKTEIYLVNKGKTI FRFSATNALVYLSPPFI 120
QY 121 RRAAVKIIVHSLENNLIMCTILTNCFVMAQNDPPMTKXVEYTFATYTFESLVKILAR 180
DB 121 RRAAVKIIVHSLENNLIMCTILTNCFVMAQNDPPMTKXVEYTFATYTFESLVKILAR 180
QY 181 FCLAAFTFLRDPNMWLDPSVIIIMAYTTEFVDLGNVSAIRTPRVBALKTIVISGLKTI 240
DB 181 FCLAAFTFLRDPNMWLDPSVIIIMAYTTEFVDLGNVSAIRTPRVBALKTIVISGLKTI 240
QY 241 GALIISVKKLADVWVLTVCISVFPALIGLQFMGNLRHKCVANFTALNGTNGSVBADGLV 300
DB 241 GALIISVKKLADVWVLTVCISVFPALIGLQFMGNLRHKCVANFTALNGTNGSVBADGLV 300
QY 301 WESLDLYSDPENNYLLKNGTSDVLLCGNSSDAGTCEGYRCLKAGENDPHGYTSFDSFAM 360
DB 301 WESLDLYSDPENNYLLKNGTSDVLLCGNSSDAGTCEGYRCLKAGENDPHGYTSFDSFAM 360
QY 361 AFLAFLRLMTODCWEELYOOTLRSAKTIYIFFMVLI FGSFYLVNLLIAVAMAYEEN 420
DB 361 AFLAFLRLMTODCWEELYOOTLRSAKTIYIFFMVLI FGSFYLVNLLIAVAMAYEEN 420
QY 421 QATIAETEKEKRFQAMEMLKKEHEALFIRGVDTYRSLSLEKSPAPVNSHERSKRR 480
DB 421 QATIAETEKEKRFQAMEMLKKEHEALFIRGVDTYRSLSLEKSPAPVNSHERSKRR 480
QY 481 RNSSGTEECGEDRLPKSDESDGPRANMHLSTRGLSRTSMKPRSSRGSIFTRRRDLGSE 540
DB 481 RNSSGTEECGEDRLPKSDESDGPRANMHLSTRGLSRTSMKPRSSRGSIFTRRRDLGSE 540
QY 541 ADPADDENSTAGESESHRTSLVPPWLRRTSAQOGSPGTSAPGALHKKNSYDPCNV 600
DB 541 ADPADDENSTAGESESHRTSLVPPWLRRTSAQOGSPGTSAPGALHKKNSYDPCNV 600
QY 601 VSLLAGDPEATSPGSHLARPVWLBNRPDTTTPSEBPGPQMLTSGAPCVDPGEERGARQ 660

DB VSLLAGDPEATSPGSHLARPVWLBNRPDTTTPSEBPGPQMLTSGAPCVDPGEERGARQ 660
QY 661 RALSAVSVLTSALEELSESRHKCPCCMNRLAOKYVLIWCCCPLMWSIKOGKLVVMPDFT 720
DB 661 RALSAVSVLTSALEELSESRHKCPCCMNRLAOKYVLIWCCCPLMWSIKOGKLVVMPDFT 720
QY 721 LTTTMCIVANTLFMALEHVNMTSEFERMLQVGNLVFTGIFTAEMTEKIALADPYYPFOG 780
DB 721 LTTTMCIVANTLFMALEHVNMTSEFERMLQVGNLVFTGIFTAEMTEKIALADPYYPFOG 780
QY 781 WNI FDSIIIVLSLMEIGLSMWSNLVSRFRLLRVFKLASWPTLNTLKIIGNSVAGL 840
DB 781 WNI FDSIIIVLSLMEIGLSMWSNLVSRFRLLRVFKLASWPTLNTLKIIGNSVAGL 840
QY 841 NLTIVLAIIVFTAVVGMOLFKNYSRLRSDSGLLPRHMMDFPHAFLIIFRLGEMV 900
DB 841 NLTIVLAIIVFTAVVGMOLFKNYSRLRSDSGLLPRHMMDFPHAFLIIFRLGEMV 900
QY 901 ETMWDCEVSGOSICLLVFLVWVIGNLVLANFLALLSSFSADNLTAPDEDERENMLQ 960
DB 901 ETMWDCEVSGOSICLLVFLVWVIGNLVLANFLALLSSFSADNLTAPDEDERENMLQ 960
QY 961 LALARIQGLRPVKRTTWDPCCGLLRORPOKPAALAAQOLPSCIATPYSPPEETKVP 1020
DB 961 LALARIQGLRPVKRTTWDPCCGLLRORPOKPAALAAQOLPSCIATPYSPPEETKVP 1020
QY 1021 PTRKETPERGEOPGCGTDPDPPVCPVIVASDPTDOBEDEENSLGTEESKQESOP 1080
DB 1021 PTRKETPERGEOPGCGTDPDPPVCPVIVASDPTDOBEDEENSLGTEESKQESOP 1080
QY 1081 VSGGEAPPDPSRTSQQVSATASSAEASASQADWRQOKAEPQAPCGGETPEDSCSEG 1140
DB 1081 VSGGEAPPDPSRTSQQVSATASSAEASASQADWRQOKAEPQAPCGGETPEDSCSEG 1140
QY 1141 ADMNTAELLBOIPDLQDYVDPEDCCTBEGCVRRCPCCAVDTTOAPBKWWRRLKTYHI 1200
DB 1141 ADMNTAELLBOIPDLQDYVDPEDCCTBEGCVRRCPCCAVDTTOAPBKWWRRLKTYHI 1200
QY 1201 VEHSMFETFIIFMLISSGALAFEDIYLBEEKITKVLLEYADKKFTVFVLEMLKKVAY 1260
DB 1201 VEHSMFETFIIFMLISSGALAFEDIYLBEEKITKVLLEYADKKFTVFVLEMLKKVAY 1260
QY 1261 GFKKYFTNAMCMTDFLIVDSVLSLVANTLGFAGMGP IKSRLTRLRALRPALSRFGMR 1320
DB 1261 GFKKYFTNAMCMTDFLIVDSVLSLVANTLGFAGMGP IKSRLTRLRALRPALSRFGMR 1320
QY 1321 VVNAALVGAIPSIINVLVCLIFMLIFSINGVNLFAKFGRCINQTEGDLPLANTIVNNK 1380
DB 1321 VVNAALVGAIPSIINVLVCLIFMLIFSINGVNLFAKFGRCINQTEGDLPLANTIVNNK 1380
QY 1381 SCSLSMLTGEIYWKVKNVDNAGAGYLLAQVATFKGMWDIYAAVDSRGYEQOWE 1440
DB 1381 SCSLSMLTGEIYWKVKNVDNAGAGYLLAQVATFKGMWDIYAAVDSRGYEQOWE 1440
QY 1441 YNLWYIYFVFIIFGSEFTLNLFGVLIIDNFNOQKKLGQDI FMTBEOKKYNNAMKL 1500
DB 1441 YNLWYIYFVFIIFGSEFTLNLFGVLIIDNFNOQKKLGQDI FMTBEOKKYNNAMKL 1500
QY 1501 GSKKPQKPIPRPLNKYQGFIDYITKQAFDYTIMFLCLANNVTMMVETDDOSP KINILA 1560
DB 1501 GSKKPQKPIPRPLNKYQGFIDYITKQAFDYTIMFLCLANNVTMMVETDDOSP KINILA 1560
QY 1561 KINILFVAIFPGECIVKLAALRHYVFNMSNVI PPVVVYIISIVGTVSDIIQKFFSPTL 1620
DB 1561 KINILFVAIFPGECIVKLAALRHYVFNMSNVI PPVVVYIISIVGTVSDIIQKFFSPTL 1620
QY 1621 FRVIRLARIIGILRLIRGAKIRTLFLAMSLPALFNIGLFLVNETYSIFGMANFAY 1680
DB 1621 FRVIRLARIIGILRLIRGAKIRTLFLAMSLPALFNIGLFLVNETYSIFGMANFAY 1680
QY 1681 VKWEAGIDMFNFQTPANSMLCLFOITTSAGMDGLSPIINTGPPYCDPTLPNSNGSGD 1740

Db 1661 VKWAGIDDMENFQTPANSMCLFQITTSAGMDGLSLINTGPPYCDPTLPNSNGSRD 1740
 Qy 1741 CGSPAVGILFTFTYIIISFLIVNNYIAIIENFSVATEESTREPLSEDDPMFYEIRKEF 1800
 Db 1741 CGSPAVGILFTFTYIIISFLIVNNYIAIIENFSVATEESTREPLSEDDPMFYEIRKEF 1800
 Qy 1801 DPEATQFIYSVLSDFADALSEPLRIAKPNOISLIMDLPMVSGDRHICMDILFAFTKRV 1860
 Db 1801 DPEATQFIYSVLSDFADALSEPLRIAKPNOISLIMDLPMVSGDRHICMDILFAFTKRV 1860
 Qy 1861 LGESGEMDALKIOMEKEMANPSKISTYEPTITTLRRGHEVSAMVIOPAFRRLQSL 1920
 Db 1861 LGESGEMDALKIOMEKEMANPSKISTYEPTITTLRRGHEVSAMVIOPAFRRLQSL 1920
 Qy 1921 KHASFLFRQAGSGISEDAPEREGLIAYVMSSENRPLGPPSSSISTSPSPSYDVT 1980
 Db 1921 KHASFLFRQAGSGISEDAPEREGLIAYVMSSENRPLGPPSSSISTSPSPSYDVT 1980
 Qy 1981 RATSNDLQVRGSDYSHSEDLADFPSPDRDRESIV 2015
 Db 1981 RATSNDLQVRGSDYSHSEDLADFPSPDRDRESIV 2015
 RESULT 7
 AEB22994 ID AEB22994 standard; protein; 2015 AA.
 AC AEB22994;
 XX 22-SEP-2005 (first entry)
 DE Mutant human SCN5A protein, G292S.
 XX
 KM SCN5A; sodium channel; Brugada syndrome; heart arrhythmia;
 KM antiarrhythmic; selectable marker; ventricular fibrillation; mutcin.
 OS Homo sapiens.
 OS Synthetic.
 OS
 FH Key Location/Qualifiers
 FT Misc-difference 292
 FT /note= "The wild-type Gly residue has been substituted
 with Ser"
 PN JP2005192413-A.
 XX 21-JUL-2005.
 PD 26-DEC-2003; 2003JP-00435234.
 PF 26-DEC-2003; 2003JP-00435234.
 PR 26-DEC-2003; 2003JP-00435234.
 XX (DOKU-) DOKURITSU GYOSEI HOJIN KAGAKU GIJUTSU SH.
 PA Matsunaga A;
 PI
 PI
 PI
 DR WPI; 2005-501993/51.
 XX
 XX The invention relates to a novel SCN5A gene of a sodium channel alpha
 CC subunit. The novel SCN5A gene comprises the mutation G292S between the
 CC fifth and sixth membrane passing-through subunits of the first domain;
 CC and/or the mutation S835L of the intracellular loop between the fourth
 CC and fifth membrane passing-through subunits of the second domain. The
 CC novel SCN5A gene can be used as a Brugada syndrome marker, for diagnosing
 CC Brugada syndrome. Brugada syndrome causes idiopathic ventricular
 CC fibrillation, thus the gene/marker allow for the prevention or treatment

CC of the syndrome. This sequence represents the G292S mutant protein of the
 CC human SCN5A gene of the invention.
 XX
 SQ Sequence 2015 AA;
 Query Match 99.9%; Score 10478; DB 9; Length 2015;
 Best Local Similarity 99.9%; Pred. No. 0;
 Matches 2013; Conservative 0; Mismatches 2; Indels 0; Gaps 0;
 Qy 1 MANFLPRTGSSPFRFPRESLAIEKMAEKOAGSTTLQSRREGLPREBA PRPDLQ 60
 Db 1 MANFLPRTGSSPFRFPRESLAIEKMAEKOAGSTTLQSRREGLPREBA PRPDLQ 60
 Qy 61 SKKLPDLXGNPQBLIGPLEDLPFYSTQKTFIVLNKGTIFRFSATNALVLSPPHPI 120
 Db 61 SKKLPDLXGNPQBLIGPLEDLPFYSTQKTFIVLNKGTIFRFSATNALVLSPPHPI 120
 Qy 121 RRAAVKILVHSLFMNLIMCTILTNCFVMAQHDPPPWTKYVEYTFATYTFESLVKILARG 180
 Db 121 RRAAVKILVHSLFMNLIMCTILTNCFVMAQHDPPPWTKYVEYTFATYTFESLVKILARG 180
 Qy 181 FCHAFPTFLRDPKMWLDPSVILMAVYTFEVDLGNVSAKRTFRVRLAKTISVISGLKTIY 240
 Db 181 FCHAFPTFLRDPKMWLDPSVILMAVYTFEVDLGNVSAKRTFRVRLAKTISVISGLKTIY 240
 Qy 241 GALIOSVVKLADVWVLTVPCLSVFALIGLOLPMGNLRRKCYRNPFTALNGTGSYEADGLV 300
 Db 241 GALIOSVVKLADVWVLTVPCLSVFALIGLOLPMGNLRRKCYRNPFTALNGTGSYEADGLV 300
 Qy 301 WESLDLYLSDPENYLNKNGTSDVLLCGNSSDAGTCPEGYRCLKAGENDGYTSFSPFAM 360
 Db 301 WESLDLYLSDPENYLNKNGTSDVLLCGNSSDAGTCPEGYRCLKAGENDGYTSFSPFAM 360
 Qy 361 AFLALFRLMTODCHERLYOQTLRSAGKITYMFMLVIFLGSFYLVNLLAVANAYEENON 420
 Db 361 AFLALFRLMTODCHERLYOQTLRSAGKITYMFMLVIFLGSFYLVNLLAVANAYEENON 420
 Qy 421 QATIAETBEKRRFOEAMEMKKEHEALITIGVTVSRSSLEMSPLAPVNSHERSKRK 480
 Db 421 QATIAETBEKRRFOEAMEMKKEHEALITIGVTVSRSSLEMSPLAPVNSHERSKRK 480
 Qy 481 RMSSGTECEGDRLPKSDSEDPGRAMNHLSTRGLSRTSMKPRSSRSGSIFFRRLDGE 540
 Db 481 RMSSGTECEGDRLPKSDSEDPGRAMNHLSTRGLSRTSMKPRSSRSGSIFFRRLDGE 540
 Qy 541 ADPADDENSTRGSESHRTSLVWPPLRRTSACQOPBGTSAFGHALHGKKNSTYDCGV 600
 Db 541 ADPADDENSTRGSESHRTSLVWPPLRRTSACQOPBGTSAFGHALHGKKNSTYDCGV 600
 Qy 601 VSLIAGDPREATSPGSHLRPVMLRHPDPTTPEBEPGQPOMLTSOAPCVUGPBERGARQ 660
 Db 601 VSLIAGDPREATSPGSHLRPVMLRHPDPTTPEBEPGQPOMLTSOAPCVUGPBERGARQ 660
 Qy 661 PALSAAVSLTSALBELESRRHKCPCKNRLAQRVLIWCCPLAMSIKQVGLVMDPPTD 720
 Db 661 PALSAAVSLTSALBELESRRHKCPCKNRLAQRVLIWCCPLAMSIKQVGLVMDPPTD 720
 Qy 721 LITIMCIIVLTLFALHYNMTSEFEEMLOVGNLVFTGIFPAENTFKIALLDPYTYFOQG 780
 Db 721 LITIMCIIVLTLFALHYNMTSEFEEMLOVGNLVFTGIFPAENTFKIALLDPYTYFOQG 780
 Qy 781 WNIPEDSIIVLSLMEIGLSWNSLTVLRSPFLARVFLAASMPILNTLTIKISVGLAG 840
 Db 781 WNIPEDSIIVLSLMEIGLSWNSLTVLRSPFLARVFLAASMPILNTLTIKISVGLAG 840
 Qy 841 NLTLVLAIVIFAVVGMOLFQKNYSRLRSDSGILPRMHMDFFAFLIIFRILCGEMI 900
 Db 841 NLTLVLAIVIFAVVGMOLFQKNYSRLRSDSGILPRMHMDFFAFLIIFRILCGEMI 900
 Qy 901 ETTMDCHEVSGQSCLLVPLLVNVTGNLVNLTFLALLSSFSADNITAPDEDEMMNLQ 960
 Db 901 ETTMDCHEVSGQSCLLVPLLVNVTGNLVNLTFLALLSSFSADNITAPDEDEMMNLQ 960

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QY 961 LALARIQGLAFVKKRTTWDFFCGILLRORPOKPAALAAQOLPSCIATPSPPEKVP 1020
DB 961 LALARIQGLAFVKKRTTWDFFCGILLRORPOKPAALAAQOLPSCIATPSPPEKVP 1020
QY 1021 PTKRETRREEEOQCGQTPGDEPCVPIAAVESDPTDDEENSLGTEBESSKOESOP 1080
DB 1021 PTKRETRREEEOQCGQTPGDEPCVPIAAVESDPTDDEENSLGTEBESSKOESOP 1080
QY 1081 VSGPEAPDSTRYMSQVATASSEASASQADMWQKMAEPQAPCGCETPEDSCSBST 1140
DB 1081 VSGPEAPDSTRYMSQVATASSEASASQADMWQKMAEPQAPCGCETPEDSCSBST 1140
QY 1141 ADMNTAELEQIPLGQDVDPEDCFTEGCVRRCPCCAVDTTQAPGVMMRLKRTCYHI 1200
DB 1141 ADMNTAELEQIPLGQDVDPEDCFTEGCVRRCPCCAVDTTQAPGVMMRLKRTCYHI 1200
QY 1201 VEHSMFEFFIIFMILLSSGALAFEDIYLBERTIKVLEVDKMFYVLEMLIKWAV 1260
DB 1201 VEHSMFEFFIIFMILLSSGALAFEDIYLBERTIKVLEVDKMFYVLEMLIKWAV 1260
QY 1261 GPKKYFTNAMCWLDFLIYDVSLVSVANTLQFAEMGPISLRTLALRPLALSRFEGMR 1320
DB 1261 GPKKYFTNAMCWLDFLIYDVSLVSVANTLQFAEMGPISLRTLALRPLALSRFEGMR 1320
QY 1321 VVNAALVGAISIMNVLLVCLIFMLIFSIMGVNLFAGKRGRCINOTEGDLPLANTYVNNK 1380
DB 1321 VVNAALVGAISIMNVLLVCLIFMLIFSIMGVNLFAGKRGRCINOTEGDLPLANTYVNNK 1380
QY 1381 SQCESLNTGELYMTKVKVNFEDNVGAGYALALQVATPFQGMNDIMYAADVSRYGEPOME 1440
DB 1381 SQCESLNTGELYMTKVKVNFEDNVGAGYALALQVATPFQGMNDIMYAADVSRYGEPOME 1440
QY 1441 YNLWYIVFVFIIFGSPFTNLPIGVIIIDNENQOKKLGGODIPMTEBOKKYTNAMKUL 1500
DB 1441 YNLWYIVFVFIIFGSPFTNLPIGVIIIDNENQOKKLGGODIPMTEBOKKYTNAMKUL 1500
QY 1501 GSKKROKPIPRPINKYOGPIPIQVTKOAFDVTIMFLICLNVTMWEVDSDSPKINTILA 1560
DB 1501 GSKKROKPIPRPINKYOGPIPIQVTKOAFDVTIMFLICLNVTMWEVDSDSPKINTILA 1560
QY 1561 KINLFLVAIFTEGECIVKLAALRHYFYFTNSWNI FDEVVVILSLVGTLSLIIQKXFSPUL 1620
DB 1561 KINLFLVAIFTEGECIVKLAALRHYFYFTNSWNI FDEVVVILSLVGTLSLIIQKXFSPUL 1620
QY 1621 FRVIRLARIGRILRLIRGAKGIRTLIPALMNSLPALFNIGLILFVMEFYISFGMANPAY 1680
DB 1621 FRVIRLARIGRILRLIRGAKGIRTLIPALMNSLPALFNIGLILFVMEFYISFGMANPAY 1680
QY 1681 VKMEGIDDMENFORFANSMCLFOITTSAGMDGLSPLINTGPPYCDPTLPNSNGSGRD 1740
DB 1681 VKMEGIDDMENFORFANSMCLFOITTSAGMDGLSPLINTGPPYCDPTLPNSNGSGRD 1740
QY 1741 CGSPAVGLFTFTYIIISFLIYVNNYIAIILENFVATEESTEPSEDDFDMFYIWEKF 1800
DB 1741 CGSPAVGLFTFTYIIISFLIYVNNYIAIILENFVATEESTEPSEDDFDMFYIWEKF 1800
QY 1801 DPEATQFI EYSVLDPADALSEPLRIAKPNQISLINMDLPVSGDRHICMDILFAFYRY 1860
DB 1801 DPEATQFI EYSVLDPADALSEPLRIAKPNQISLINMDLPVSGDRHICMDILFAFYRY 1860
QY 1861 LGSESEMALKIOMEKEMPAANPSKISYEPIITTLRRHEEVSANVIOQAPRRHLOSL 1920
DB 1861 LGSESEMALKIOMEKEMPAANPSKISYEPIITTLRRHEEVSANVIOQAPRRHLOSL 1920
QY 1921 KHAFLFRQAGSGSEEDAPAREGLIAYVMSSENFSPGPPSSSISISTSPSPSYDVT 1980
DB 1921 KHAFLFRQAGSGSEEDAPAREGLIAYVMSSENFSPGPPSSSISISTSPSPSYDVT 1980
QY 1981 RATSNDNLQVRGSDYSHSBDLADPPSPDRRESIV 2015
DB 1981 RATSNDNLQVRGSDYSHSBDLADPPSPDRRESIV 2015

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RESULT 8
ADM33995
ID ADM33995 standard; protein: 2016 AA.
XX
AC ADM33995;
XX
DT 03-JUN-2004 (first entry)
XX
DE Human SCN5A variant 1 protein SEQ ID NO:2.
XX
KW human; cardiac sodium channel alpha subunit; SCN5A;
KW sodium voltage-gated channel; type V; alpha; mutation study;
KW sodium channel related disease.
XX
OS Homo sapiens.
XX
MO M02004012668-A2.
XX
PD 12-FEB-2004.
XX
PE 01-AUG-2003; 2003WO-US024190.
XX
PR 02-AUG-2002; 2002US-0401018P.
XX
PA (WISC ) WISCONSIN ALUMNI RES FOUND.
XX
PI Makiejski JC, Ye B, Ackerman MJ,
XX
DR WPI; 2004-157000/15.
XX
DR N-PSDB; ADM33994.
XX
PT New nucleic acid encoding sodium voltage-gated channel, type V, alpha
PT polypeptide variants, useful for studying mutations, and designing or
PT identifying new diagnostics and treatment strategies or agents for sodium
PT channel related diseases.
XX
PS Claim 8; SEQ ID NO 2; 11pp; English.
XX
CC The present sequence represents a variant of the human cardiac sodium
CC channel alpha subunit designated SCN5A (sodium voltage-gated channel,
CC type V, alpha) protein. The present invention describes an isolated
CC polynucleotide encoding an SCN5A polypeptide where the polynucleotide is
CC selected from the group: (1) a first polynucleotide that encodes an SCN5A
CC polypeptide selected from the group consisting of: (i) a histidine,
CC threonine, leucine, arginine and glutamine at amino acid positions 558,
CC 559, 618, 1027 and 1077, respectively; (ii) an arginine, threonine,
CC 559, 618, 1027 and 1077, respectively; (iii) a histidine, threonine, leucine and
CC 1027 and 1077, respectively; (iv) a histidine, threonine, leucine and
CC arginine at amino acid positions 558, 559, 618 and 1027, respectively,
CC with the amino acid at amino acid position 1077 deleted; or (iv) an
CC arginine, threonine, leucine and arginine at amino acid positions 558,
CC 559, 618 and 1027, respectively, with the amino acid at amino acid
CC position 1077 deleted; (2) a second polynucleotide that is at least 80 %
CC identical to the first polynucleotide over the entire length of the first
CC polypeptide; (3) a third polynucleotide that encodes any of the SCN5A
CC polypeptides with a conservative substitution, deletion or rearrangement
CC at one or more non-critical amino acid position; and (4) a fourth
CC polynucleotide that is a complement of the first, second or third
CC polynucleotide. Also described: (1) a genetic construct comprising the
CC polynucleotide operably linked to a non-native expression control
CC sequence; (2) a cell comprising the polynucleotide; (3) an isolated
CC polypeptide encoded by the polynucleotide; (4) an antibody that
CC specifically binds to the polypeptide; (5) identifying an agent that can
CC alter the activity of a sodium channel; (6) identifying an agent that can
CC alter the expression of a sodium channel; (7) determining whether a
CC biological sample or a preparation derived from the biological sample
CC contains the polypeptide; (8) determining whether a mutation on a sodium
CC channel is associated with a disease; and (9) determining whether a human
CC or non-human subject is at risk for Long QT syndrome. The SCN5A
CC polynucleotides and polypeptides are useful for studying mutations, and
CC designing or identifying new diagnostics and treatment strategies or
CC agents for sodium channel related diseases or conditions.
XX

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SQ Sequence 2016 AA:	
Query Match	99.9%; Score 10473.5; DB 8; Length 2016;
Beet Local Similarity	99.9%; Pred. No. 0;
Matches 2014; Conservative	0; Mismatches 1; Indels 1; Gaps 1;
QY	1 MANPLPGTSTFRRTRESLAIAEKMAEKQARGSTTLQESREGLPREEAPRPOLDQA 60
DB	1 MANPLPGTSTFRRTRESLAIAEKMAEKQARGSTTLQESREGLPREEAPRPOLDQA 60
QY	61 SKKPDLYGNPPQSLIGEPLEDLPDYSTQKTFIVLANKGKTIFFRSATNALVYLSPPHI 120
DB	61 SKKPDLYGNPPQSLIGEPLEDLPDYSTQKTFIVLANKGKTIFFRSATNALVYLSPPHI 120
QY	121 RRAAKIIIVHSLENNLIMCTIITNCVMAQHPDPWTQVETTFATITTESLVKILARG 180
DB	121 RRAAKIIIVHSLENNLIMCTIITNCVMAQHPDPWTQVETTFATITTESLVKILARG 180
QY	181 FCLHAFTELDPNMWLDPSVIIIMAYTTEFVDLGANVSALRTFRVLKALXTISVIGLAKTIV 240
DB	181 FCLHAFTELDPNMWLDPSVIIIMAYTTEFVDLGANVSALRTFRVLKALXTISVIGLAKTIV 240
QY	241 GALIOSVKKADAVMYLTVFCISVPAIIIGLQFMGNLRHKCVRNFTALNGTNGSVADGLV 300
DB	241 GALIOSVKKADAVMYLTVFCISVPAIIIGLQFMGNLRHKCVRNFTALNGTNGSVADGLV 300
QY	301 MESIDLVIYSDPENYLLKNGTSDVLLCGNSSDAGTCPEGYRCIKAGENDHGTSTFDSFAM 360
DB	301 MESIDLVIYSDPENYLLKNGTSDVLLCGNSSDAGTCPEGYRCIKAGENDHGTSTFDSFAM 360
QY	361 AFLALFRIMTODCMERLYQOTLRSAKITYMIFFMVLVIFLGSFYLVNLIIVAVMAAYEON 420
DB	361 AFLALFRIMTODCMERLYQOTLRSAKITYMIFFMVLVIFLGSFYLVNLIIVAVMAAYEON 420
QY	421 QATIAETEKEKRFQEAEMELKKEHEALTIRGVDTVSSSLEMSDPLAVNSHERSKKRX 480
DB	421 QATIAETEKEKRFQEAEMELKKEHEALTIRGVDTVSSSLEMSDPLAVNSHERSKKRX 480
QY	481 RMSSGTECGEDRLPKSDESDGPRAMNLSITRGLSRTSMKPRSRGSIIFFRRDDGSE 540
DB	481 RMSSGTECGEDRLPKSDESDGPRAMNLSITRGLSRTSMKPRSRGSIIFFRRDDGSE 540
QY	541 ADPADENSTAGESESHRTSLVPMPLRTSAQOGPSPTGAPGALHGGKNSYDPCGV 600
DB	541 ADPADENSTAGESESHRTSLVPMPLRTSAQOGPSPTGAPGALHGGKNSYDPCGV 600
QY	601 VSLIAGDPEATSPGSHLLRPVMLEHPDITTPSEBPGPOMLTSAQPCVDGFEBPGARQ 660
DB	601 VSLIAGDPEATSPGSHLLRPVMLEHPDITTPSEBPGPOMLTSAQPCVDGFEBPGARQ 660
QY	661 RALSAVSULTSALBELBSRHKCPCCNRILAORYIIMCCPLMNSIKQGVKLVMDPPTD 720
DB	661 RALSAVSULTSALBELBSRHKCPCCNRILAORYIIMCCPLMNSIKQGVKLVMDPPTD 720
QY	721 LTTIMCIYANTLFMALBHYNMTSEFEEMLYQGNLVFTGIIFTAEMTFKIIALDPYYFQGG 780
DB	721 LTTIMCIYANTLFMALBHYNMTSEFEEMLYQGNLVFTGIIFTAEMTFKIIALDPYYFQGG 780
QY	781 WNFDSIIIVILSMELGSRMSNLSVLSFRLLRVFKLAKSPMLTILIKIIGNSVGLG 840
DB	781 WNFDSIIIVILSMELGSRMSNLSVLSFRLLRVFKLAKSPMLTILIKIIGNSVGLG 840
QY	841 NLTLVLAIVFIYAVNGMQLFGKNYSLEIRSDSGLLPRMNMDFPHAFLLIFRILCGMI 900
DB	841 NLTLVLAIVFIYAVNGMQLFGKNYSLEIRSDSGLLPRMNMDFPHAFLLIFRILCGMI 900
QY	901 ETMMDCEVSGQSCLVFLVMTYGNLVNLFATALLSSFSADNTJAPDEBEMNLQ 960
DB	901 ETMMDCEVSGQSCLVFLVMTYGNLVNLFATALLSSFSADNTJAPDEBEMNLQ 960
QY	961 LALARIOGLRFVRRITWDFCCGLLRORPQKPAALAAQGLPSCIATPSPPPETEKVP 1020
DB	961 LALARIOGLRFVRRITWDFCCGLLRORPQKPAALAAQGLPSCIATPSPPPETEKVP 1020

QY	1021 PTRKETREBEGOPQOGTGPDPBYCVPIAAASPTDDOEDEENSLGTEBESSK-ORSQ 1079
DB	1021 PTRKETREBEGOPQOGTGPDPBYCVPIAAASPTDDOEDEENSLGTEBESSK-ORSQ 1080
QY	1080 PVSQGPAPDPSRTWSQVSATASSSEABASAOADNRQOMKPEQAPGCGETPEDSCBSG 1139
DB	1081 PVSQGPAPDPSRTWSQVSATASSSEABASAOADNRQOMKPEQAPGCGETPEDSCBSG 1140
QY	1140 TADMTNTALLBOIPDLQDYKDPEDCTBECVRRCPCCAVDTTQAPGKWMRLAKTCYH 1199
DB	1141 TADMTNTALLBOIPDLQDYKDPEDCTBECVRRCPCCAVDTTQAPGKWMRLAKTCYH 1200
QY	1200 IVESHMEFTFIIIFMLISSGALAPEDIYLEBRKTIKYLEVADKMFYVFLVEMLLKNYA 1259
DB	1201 IVESHMEFTFIIIFMLISSGALAPEDIYLEBRKTIKYLEVADKMFYVFLVEMLLKNYA 1260
QY	1260 YGPKKYFTNAMCWLDFLIVDSVLSVANTLGAFEMGPIKSLRTLRLRPLRALSREFGM 1319
DB	1261 YGPKKYFTNAMCWLDFLIVDSVLSVANTLGAFEMGPIKSLRTLRLRPLRALSREFGM 1320
QY	1320 RYVYNALVGAIPSTIMNVLVCLIFWLIFSINGVNLPAKRGRCINQTEGDLPLANTYNN 1379
DB	1321 RYVYNALVGAIPSTIMNVLVCLIFWLIFSINGVNLPAKRGRCINQTEGDLPLANTYNN 1380
QY	1380 KSGCESLNLGELVMTKVNPDNVGAGYLALQVAFKGMMDIMYAAVDSRGYEEOPOM 1439
DB	1381 KSGCESLNLGELVMTKVNPDNVGAGYLALQVAFKGMMDIMYAAVDSRGYEEOPOM 1440
QY	1440 EYNLYMYIYEFIIIFGSFPTLANFIGVINDFNQKKLGGODIFMTEBOKKYANAMK 1499
DB	1441 EYNLYMYIYEFIIIFGSFPTLANFIGVINDFNQKKLGGODIFMTEBOKKYANAMK 1500
QY	1500 LGSKKPOKPIPRPLNKOQGFIEDIVTKOAPDVTIMFLICLNMVTMVEETDQSPKINIL 1559
DB	1501 LGSKKPOKPIPRPLNKOQGFIEDIVTKOAPDVTIMFLICLNMVTMVEETDQSPKINIL 1560
QY	1560 AKINLLFVAIPTEGCIYKLAALRHYFTNSNINIDPVVILISYGVLSDIIOYFESPT 1619
DB	1561 AKINLLFVAIPTEGCIYKLAALRHYFTNSNINIDPVVILISYGVLSDIIOYFESPT 1620
QY	1620 LFRVIRIARIGRIILRLIRGAKGIRTLFPALMMSLPALFNIGLLFLVMFIYSIFGMANFA 1679
DB	1621 LFRVIRIARIGRIILRLIRGAKGIRTLFPALMMSLPALFNIGLLFLVMFIYSIFGMANFA 1680
QY	1680 YVKMEAGIDMFWNFQTPANSMLCLFOITTSAGMDGLSPILANTGPYCDPTLPNSNGSRG 1739
DB	1681 YVKMEAGIDMFWNFQTPANSMLCLFOITTSAGMDGLSPILANTGPYCDPTLPNSNGSRG 1740
QY	1740 DCGSPANGILFETTYIIISFLIVNMATIIILENFVATEBTEPSEDDDMYEIWEK 1799
DB	1741 DCGSPANGILFETTYIIISFLIVNMATIIILENFVATEBTEPSEDDDMYEIWEK 1800
QY	1800 FDPAPTOFIEYSVLSDPADLSEPLRIAKPNQISLIMNDLPMVSGDIRIHCMDIIPAFYKR 1859
DB	1801 FDPAPTOFIEYSVLSDPADLSEPLRIAKPNQISLIMNDLPMVSGDIRIHCMDIIPAFYKR 1860
QY	1860 VLGESEMDALIKIOMEKEFMAANPSKISYEDITTTLRKRIEVSAMVIQRAFRHLLORS 1919
DB	1861 VLGESEMDALIKIOMEKEFMAANPSKISYEDITTTLRKRIEVSAMVIQRAFRHLLORS 1920
QY	1920 LKXASFLFRQOAGSGLEBDAPEBEGIIAIVMSNFSRPLGPPSSSISSTSPPSYDSV 1979
DB	1921 LKXASFLFRQOAGSGLEBDAPEBEGIIAIVMSNFSRPLGPPSSSISSTSPPSYDSV 1980
QY	1980 TRATSNDLVQRGSDYSHSEDLADFPSPDPDRRESIV 2015
DB	1981 TRATSNDLVQRGSDYSHSEDLADFPSPDPDRRESIV 2016

RESULT 9
AEF90518
ID AEF90518 standard; protein; 2016 AA.

XX AEF90518;
AC 20-APR-2006 (first entry)
XX Human SCNS splice variant 1 SEQ ID NO 68.
DE antiarrhythmic; gene therapy; screening; diagnosis; prognosis;
XX SNP detection; genetic marker; sudden infant death syndrome; SCNS;
XX voltage-gated sodium channel.
OS Homo sapiens.
XX WO2006019984-A2.
XX 23-FEB-2006.
XX 15-JUL-2005; 2005WO-US025099.
XX 15-JUL-2004; 2004US-0588302P.
XX 14-APR-2005; 500US-05546987.
XX (UYVA) UNIV YALE.
XX Goldstein SAN, Bowers PN;
XX WPI; 2006-174087/18.
DR N-PSDB; AEF90517.
PT Testing a human individual for a marker for Sudden Infant Death Syndrome
PT comprises identifying a polymorphism in the cardiac sodium channel SCNSA
PT protein.
XX Claim 3; SEQ ID NO 68; 88bp; English.
XX The invention describes a method of testing a human individual for a
XX marker for Sudden Infant Death Syndrome (SIDS) comprising identifying a
XX polymorphism in a nucleic acid sample from the individual, where the
XX sample comprises both allelic copies of a nucleic acid encoding an SCNSA
XX polypeptide or copies of a protein encoded by the individual's SCNSA
XX nucleic acid. Also described are: a method of assessing a risk for SIDS
XX in a human individual; a method of identifying a human carrier of a
XX marker for SIDS; a kit, for testing a human infant or fetus for a marker,
XX assessing a risk, or for identifying a human carrier of a marker for a
XX marker for SIDS, the kit comprising: at least one reagent for identifying
XX an amino acid at a position corresponding to position 1103 in SEQ ID NO:
XX 68; and instructional material, where if the amino acid at a position
XX corresponding to position 1103 in SEQ ID NO: 2 is Y/Y, then the infant or
XX fetus has a marker for SIDS or is assessed to be at increased risk for
XX SIDS relative to an infant or fetus in which the amino acid at a position
XX corresponding to position 1103 in SEQ ID NO:68 is not Y/Y, or where if
XX the amino acid at a position corresponding to position 1103 in SEQ ID NO:
XX 68 is Y/- or Y/Y, then the human is identified as a carrier of a marker
XX for SIDS; a method of screening for SIDS in a human infant or fetus; a
XX method for diagnosing SIDS in an infant or fetus; and a method for
XX preventing SIDS in a human infant or fetus. The methods and kits are
XX useful for testing a human individual for a marker for SIDS, assessing a
XX risk for SIDS in a human individual, identifying a human carrier of a
XX marker for SIDS, and for screening and diagnosing SIDS in a human infant
XX or fetus. The method and composition are useful for preventing SIDS in a
XX human infant or fetus. This is the amino acid sequence of human SCNS
XX splice variant 1.
XX Sequence 2016 AA;
XX
XX Query Match 99.9%; Score 10473.5; DB 10; Length 2016;
XX Best Local Similarity 99.9%; Pred No. 0;
XX Matches 2014; Conservative 0; Mismatches 1; Indels 1; Gaps 1;

QY 1 MANFLPRTGTSFRFTRESLAIEKRWAEKQARGSTTLQESREGLPBEBARPLQDQA 60
DB 1 MANFLPRTGTSFRFTRESLAIEKRWAEKQARGSTTLQESREGLPBEBARPLQDQA 60

QY SKKLDPDLYGNPPOELIGEPLLEDLPFFYSTOKTFIVLNGKGTIPFASATNALVYLSPPHPI 120
DB SKKLDPDLYGNPPOELIGEPLLEDLPFFYSTOKTFIVLNGKGTIPFASATNALVYLSPPHPI 120
QY RRAAVKILVSLFNMILMCTILTNVCMAGHDPBPMTKYVEYFTALYTESLVKILARG 180
DB RRAAVKILVSLFNMILMCTILTNVCMAGHDPBPMTKYVEYFTALYTESLVKILARG 180
QY PCLHAFPLFLADPMMWLDPSYITIMAYTTEFPDLGNVSLRTPRYVALKTSIVLSGLKTVI 240
DB PCLHAFPLFLADPMMWLDPSYITIMAYTTEFPDLGNVSLRTPRYVALKTSIVLSGLKTVI 240
QY GALIQSVKCLADVWVLTFCISVPALIGLQFMGNLHKCVRNFTALNGTNGSVEADGLV 300
DB GALIQSVKCLADVWVLTFCISVPALIGLQFMGNLHKCVRNFTALNGTNGSVEADGLV 300
QY WESIDLVLSDPENYLLKNGTSDVLLCGNSSDAGTCPEGRYCLKAGENPDHGYTSFDSFAW 360
DB WESIDLVLSDPENYLLKNGTSDVLLCGNSSDAGTCPEGRYCLKAGENPDHGYTSFDSFAW 360
QY AFLALFRLMTQDCWERYLQOQLRSAGKIYVIFMLVIFLGSFYLVNLTILAVMAAYEON 420
DB AFLALFRLMTQDCWERYLQOQLRSAGKIYVIFMLVIFLGSFYLVNLTILAVMAAYEON 420
QY QATIAETEKEKRFQEMEMLKKEHEALTIRGVDTVRSLSLEMSPLAPVNSHERSRKRR 480
DB QATIAETEKEKRFQEMEMLKKEHEALTIRGVDTVRSLSLEMSPLAPVNSHERSRKRR 480
QY RMSSGTECEGDRLPKSDSEDPAMNHLSTRLGSLSTSMKPRSSRSISFTFRRDIGSE 540
DB RMSSGTECEGDRLPKSDSEDPAMNHLSTRLGSLSTSMKPRSSRSISFTFRRDIGSE 540
QY ADPADDENSTAGESESRSTSLVWPRLRRTSAQCPBPGRSAPGHALHGKKNSTVDCNGV 600
DB ADPADDENSTAGESESRSTSLVWPRLRRTSAQCPBPGRSAPGHALHGKKNSTVDCNGV 600
QY VSLGAGDPPEATSGSHLTPVMLEHPPDTTTPBEEGQPMQLTQA PCVDGFEPPARQ 660
DB VSLGAGDPPEATSGSHLTPVMLEHPPDTTTPBEEGQPMQLTQA PCVDGFEPPARQ 660
QY RALGAVSLTSALELEESRRHKCPCCNRLAQRYLWECPLMWSIKQVKLVVMDPPTD 720
DB RALGAVSLTSALELEESRRHKCPCCNRLAQRYLWECPLMWSIKQVKLVVMDPPTD 720
QY LTTMCIIVLNTFMALSHYNTSFEEMLOVGNLVFGITFAETFKIILADPYVYQQG 780
DB LTTMCIIVLNTFMALSHYNTSFEEMLOVGNLVFGITFAETFKIILADPYVYQQG 780
QY WNIPIISIVILSMELGSRMSNLVLRSPRLRVLVFKLAKSWPTLNTLIKIGNSVGALG 840
DB WNIPIISIVILSMELGSRMSNLVLRSPRLRVLVFKLAKSWPTLNTLIKIGNSVGALG 840
QY NLTVLAIIVIEFVAVGMQLFGKNYSILRSDSGLLFRWMMDFEHAFLIIFRILGEMI 900
DB NLTVLAIIVIEFVAVGMQLFGKNYSILRSDSGLLFRWMMDFEHAFLIIFRILGEMI 900
QY ETTMWDCHVEVSQSLCLVFLVWYVGNLVVNLFLALLSFSADNLTAPDEEMNLQ 960
DB ETTMWDCHVEVSQSLCLVFLVWYVGNLVVNLFLALLSFSADNLTAPDEEMNLQ 960
QY IALARIQGLRFVRRITWDFCCGLLRQRPQKRALAAGOLPSCIATPYSPPETEKVP 1020
DB IALARIQGLRFVRRITWDFCCGLLRQRPQKRALAAGOLPSCIATPYSPPETEKVP 1020
QY PTRKETRFEBGBOPOGOTPGDPEPVCPVIAVESDTDOEBEENSIGTEBESSK-OESQ 1079
DB PTRKETRFEBGBOPOGOTPGDPEPVCPVIAVESDTDOEBEENSIGTEBESSK-OESQ 1080
QY PVSQGPPEAPPSRRTWSQVSATASSEABASASQADWRQWRAEPAPPCGCFEPBSCSGS 1139
DB PVSQGPPEAPPSRRTWSQVSATASSEABASASQADWRQWRAEPAPPCGCFEPBSCSGS 1140
QY TADMTNTNALLBQIPDLGQDVKDPEDCFTEGCVARPCCAVDTTQAGKVMWRJRKTCYH 1199
DB TADMTNTNALLBQIPDLGQDVKDPEDCFTEGCVARPCCAVDTTQAGKVMWRJRKTCYH 1199

Db 1141 TADMTNTAALBQIPDLGQDVADPEDCFEGCVRBCPCCAVDTTQAPGKVMRLKRTCH 1200
 QY 1200 IVEHSMFEFTIIIMLSSGALAEPDIYEERKTKVLEVDKMTYVPELEMLKVA 1259
 Db 1201 IVEHSMFEFTIIIMLSSGALAFEDDIYEERKTKVLEVDKMTYVPELEMLKVA 1260
 QY 1260 YGFKKYFTNACMLDFLIVDSVSVANTLGAEMGPISKSLRTLRALRPLARSRFEGM 1319
 Db 1261 YGFKKYFTNACMLDFLIVDSVSVANTLGAEMGPISKSLRTLRALRPLARSRFEGM 1320
 QY 1320 RVVNAVALGAIPIISINVLVCLIFMLIPISIMGVNLPAKFGKFCINOTEGDLPLANTYVNN 1379
 Db 1321 RVVNAVALGAIPIISINVLVCLIFMLIPISIMGVNLPAKFGKFCINOTEGDLPLANTYVNN 1380
 QY 1380 KSQCSLINTGELVMTKVVNDVAGAGTALLQVATFGKMDIMYAANDSRGIEQOPM 1439
 Db 1381 KSQCSLINTGELVMTKVVNDVAGAGTALLQVATFGKMDIMYAANDSRGIEQOPM 1440
 QY 1440 EYNLYMYTYEFTIIFGSPFTLNLFIGVINDPNQOKKLAGGQDIFMTEBOKKYANAMK 1499
 Db 1441 EYNLYMYTYEFTIIFGSPFTLNLFIGVINDPNQOKKLAGGQDIFMTEBOKKYANAMK 1500
 QY 1500 LGSKKPQKPIPRPLNKYGFIDIVYKQAFDVTIMFLICLANNVTMVEITDQSPKINIL 1559
 Db 1501 LGSKKPQKPIPRPLNKYGFIDIVYKQAFDVTIMFLICLANNVTMVEITDQSPKINIL 1560
 QY 1560 AKINILFVALFTGECIVKLAALRHYYFTNSMNIPEFVVYIISIGTVLSDIIOKYFSPPT 1619
 Db 1561 AKINILFVALFTGECIVKLAALRHYYFTNSMNIPEFVVYIISIGTVLSDIIOKYFSPPT 1620
 QY 1620 LFRVRLARIGRIILIRGAKIRTLPLMMSLPLAFNIGLLPLVMFYISIFMANPA 1679
 Db 1621 LFRVRLARIGRIILIRGAKIRTLPLMMSLPLAFNIGLLPLVMFYISIFMANPA 1680
 QY 1680 YVKWAGIDDMENFOTFANSMCLFOITTSAGMDGLSPIANTGPYCDPTLPNSNGSRG 1739
 Db 1681 YVKWAGIDDMENFOTFANSMCLFOITTSAGMDGLSPIANTGPYCDPTLPNSNGSRG 1740
 QY 1740 DCGSAVGLLEFTYIIISFLIVNMVYIAIILENFSVATESTEPLSEDDPMFEIWEK 1799
 Db 1741 DCGSAVGLLEFTYIIISFLIVNMVYIAIILENFSVATESTEPLSEDDPMFEIWEK 1800
 QY 1800 FDPBATORFESVUSDPAALSEPIRIKPNQISILNMDLPMVSGDRHCHMDILPAFKR 1859
 Db 1801 FDPBATORFESVUSDPAALSEPIRIKPNQISILNMDLPMVSGDRHCHMDILPAFKR 1860
 QY 1860 VLGESEMDALAKIOMEKFMANPSKISYEPIITTLRRKHEVSAMVIOARFRHLORS 1919
 Db 1861 VLGESEMDALAKIOMEKFMANPSKISYEPIITTLRRKHEVSAMVIOARFRHLORS 1920
 QY 1920 LKHAFLFRQOAGSGLSEBDAERBGLIAYVNSENFSRPLGPPSSSISSTSPSYDSV 1979
 Db 1921 LKHAFLFRQOAGSGLSEBDAERBGLIAYVNSENFSRPLGPPSSSISSTSPSYDSV 1980
 QY 1980 TRATSDNLOVGRSDYSHSEDLADFPSPDRPRESTIV 2015
 Db 1981 TRATSDNLOVGRSDYSHSEDLADFPSPDRPRESTIV 2016

RESULT 10

AEA78664
ID AEA78664 standard; protein; 2015 AA.

AC AEA78664;

DT 25-AUG-2005 (first entry)

DE Human SCNSA sodium channel subunit (wild-type), SEQ ID NO:3.

KW Diagnoses; cardiovascular disease; heart arrhythmia; long QT syndrome;
 short QT syndrome; Brugada syndrome; progressive conduction disease;
 cardiac arrest; sodium channel, voltage-gated, type V, alpha; SCNSA;

KW single amino acid polymorphism; SNP.
 XX Homo sapiens.
 OS
 FH Key
 FT Misc-difference 104
 FT /note= "Optionally, Trp replaces wild-type Arg in a mutant SCNSA protein of the invention. Mutation is associated with Brugada syndrome"
 FT 179
 FT /note= "Optionally, an in-frame stop codon replaces wild-type Arg in a mutant SCNSA protein of the invention. Mutation is associated with Brugada syndrome"
 FT 220
 FT /note= "Optionally, Ile replaces wild-type Thr in a mutant SCNSA protein of the invention. Mutation is associated with Brugada syndrome"
 FT 232
 FT /note= "Optionally, Ile replaces wild-type Val in conjunction with the amino acid substitution L1307F in a mutant SCNSA protein of the invention. Mutation is associated with Brugada syndrome"
 FT 336
 FT /note= "Optionally, Leu replaces wild-type Pro in conjunction with the amino acid substitution I1659V in a mutant SCNSA protein of the invention. Mutation is associated with Brugada syndrome"
 FT 400
 FT /note= "Optionally, Ala replaces wild-type Gly in a mutant SCNSA protein of the invention. Mutation is associated with Brugada syndrome"
 FT 446
 FT /note= "Optionally, Lys replaces wild-type Glu in a mutant SCNSA protein of the invention. Mutation is associated with Brugada syndrome"
 FT 532
 FT /note= "Optionally, Cys replaces wild-type Phe in a mutant SCNSA protein of the invention. Mutation is associated with Brugada syndrome"
 FT 735
 FT /note= "Optionally, Val replaces wild-type Ala in a mutant SCNSA protein of the invention. Mutation is associated with Brugada syndrome"
 FT 851. 2015
 FT /note= "Optionally, this segment is replaced with the variant C-terminus CSSLLMWACSSIAKTRIS in a mutant SCNSA protein of the invention. Mutation is associated with Brugada syndrome and results from a 2 base insertion between codons 850 and 851 (T3851 mutation)"
 FT 878
 FT /note= "Optionally, Cys replaces wild-type Arg in a mutant SCNSA protein of the invention. Mutation is associated with Brugada syndrome"
 FT 886
 FT /note= "Optionally, Pro replaces wild-type His in a mutant SCNSA protein of the invention. Mutation is associated with Brugada syndrome"
 FT 917
 FT /note= "Optionally, Arg replaces wild-type Leu in a mutant SCNSA protein of the invention. Mutation is associated with Brugada syndrome"
 FT 1008
 FT /note= "Optionally, Ser replaces wild-type Pro in a mutant SCNSA protein of the invention. Mutation is associated with progressive conduction disease"
 FT 1134
 FT /note= "Optionally, Ile replaces wild-type Ser in a mutant SCNSA protein of the invention. Mutation is associated with long QT syndrome"
 FT 1307
 FT /note= "Optionally, Phe replaces wild-type Leu in conjunction with the amino acid substitution V232I in a mutant SCNSA protein of the invention. Mutation is

FT associated with Brugada syndrome"
FT 1573. .1604
FT /note= "Optionally, this segment is deleted in a mutant
FT SCN5A protein of the invention. Mutation is associated
FT with Brugada syndrome and results from aberrant splicing
FT of exons 27 and 28"
FT
FT Misc-difference 1573
FT /note= "Optionally, Lys replaces wild-type Glu in a
FT mutant SCN5A protein of the invention. Mutation is
FT associated with Brugada syndrome"
FT
FT Misc-difference 1614
FT /note= "Optionally, an in-frame stop codon replaces wild-
FT type Tyr in a mutant SCN5A protein of the invention.
FT Mutation is associated with Brugada syndrome"
FT
FT Misc-difference 1659
FT /note= "Optionally, Val replaces wild-type Ile in
FT conjunction with the amino acid substitution P336L in a
FT mutant SCN5A protein of the invention. Mutation is
FT associated with Brugada syndrome"
FT
FT Misc-difference 1727
FT /note= "Optionally, Arg replaces wild-type Cys in a
FT mutant SCN5A protein of the invention. Mutation is
FT associated with Brugada syndrome"
FT
FT US2005130190-A1.
FT
FT 16-JUN-2005.
FT
FT 23-AUG-2004; 2004US-00924375.
FT
FT 22-AUG-2003; 2003US-0497256P.
FT
FT (ANTZ/) ANTZELEVITCH C.
FT (BRUG/) BRUGADA R.
FT (HONG/) HONG K.
FT
FT Antzelevitch C, Brugada R, Hong K;
FT WPI; 2005-443796/45.
FT DR N-PSDB; AEA78665.
FT
FT Novel isolated mutant ion channel protein e.g. KCNH2 protein,
FT corresponding to wild-type human KCNH2 protein, useful for diagnosing and
FT screening drugs to treat long and short QT syndrome.
FT
FT Claim 7; SEQ ID NO 3; 65pp; English.
FT
FT The invention relates to mutants of the ion channel proteins KCNQ1
FT (potassium voltage-gated channel, KQT-like subfamily, member 1;
FT AEA78662), SCN5A (sodium channel, voltage-gated, type V, alpha; AEA78664)
FT and KCNH2 (potassium voltage-gated channel, subfamily H, member 2, also
FT known as HERG; AEA78666), and to mutant KCNQ1, SCN5A and KCNH2 nucleic
FT acids. The invention also relates to vectors and host cells comprising
FT mutant nucleic acids of the invention; nucleic acid probes which
FT selectively hybridize to a mutant KCNQ1, SCN5A or KCNH2 nucleic acid but
FT not to the corresponding wild-type polynucleotide; microarrays containing
FT such probes; and antibodies immunospecific for mutant KCNQ1, SCN5A or
FT KCNH2 proteins. The mutant proteins of the invention result from
FT previously unknown mutations in the KCNQ1, SCN5A and KCNH2 genes and are
FT involved in ion channel disruptions associated with diseases resulting in
FT arrhythmias and/or sudden cardiac death (cardiac arrest) such as short QT
FT syndrome, long QT syndrome, Brugada syndrome and progressive conduction
FT disease. The novel KCNQ1 mutation, G189W (sic), is associated with long
FT QT syndrome. Most of the novel SCN5A mutations are associated with
FT Brugada syndrome; these mutations are R104W, R179stop, T220I, G400A,
FT E44K, P53C, A735V, R878C, H886P, L917R, E1573K, Y1614stop, C1727R,
FT V2321+I1307F, P336L+I1659V, T885I (a TG base insertion between codons
FT 850 and 851 of the coding sequence), and a deletion of E1573-G1604
FT (resulting from an aberrant "splice of exons 27 and 28-4810+7 ins GGG").
FT The other novel SCN5A mutations, P1008S and S1134I are respectively
FT associated with progressive conduction disease and long QT syndrome.
FT There are 4 novel mutations of KCNH2, one of these, N588K is associated
FT with short QT syndrome, while the other three, R356H, W396stop and C764

CC deletion (a deletion of cytosine from codon 764 of the coding sequence),
CC are associated with long QT syndrome. The mutant ion channel proteins, QT
CC polynucleotides, antibodies and probes are useful for diagnosing short
CC syndrome, Brugada syndrome, progressive conduction disease or long QT
CC syndrome. They can be used to determine an individual's susceptibility to
CC sudden cardiac death, or to determine whether this is the cause of death
CC for a deceased individual. The proteins and polynucleotides are also
CC useful in screening for compounds useful in treating the above-mentioned
CC diseases. The present sequence represents the wild-type human SCN5A
CC protein.
CC
CC
XX
SQ Sequence 2015 AA;
Query Match 99.8%; Score 10471; DB 9; Length 2015;
Best Local Similarity 99.9%; Pred. No. 0;
Matches 2012; Conservative 0; Mismatches 3; Indels 0; Gaps 0;
OY 1 MANFLPRGTSSFRFRFRESIAAEKMAEKORAGSTTLQESREGLPDEEAPRPOLDQA 60
DB 1 MANFLPRGTSSFRFRFRESIAAEKMAEKORAGSTTLQESREGLPDEEAPRPOLDQA 60
OY 61 SKKLPLDLYGNPPOELICEPIEDDDPFYSTOKTFIVLANKKKTIRFSATNLVYLSPPHI 120
DB 61 SKKLPLDLYGNPPOELICEPIEDDDPFYSTOKTFIVLANKKKTIRFSATNLVYLSPPHI 120
OY 121 PRAAVKLVNSIFRPMILMCTILNVCVMAQHDPPMTKYEYPTAIYFESLVKILARG 180
DB 121 PRAAVKLVNSIFRPMILMCTILNVCVMAQHDPPMTKYEYPTAIYFESLVKILARG 180
OY 181 FCLHAFPLRDPKMWMLDFSVIIMAYTTEFVDLGNVSLARTFVRLAKTISVIGLKTIV 240
DB 181 FCLHAFPLRDPKMWMLDFSVIIMAYTTEFVDLGNVSLARTFVRLAKTISVIGLKTIV 240
OY 241 GALIQSVKCLADVAVLTTFCLSVFALIGLOFMGNLHKCVRNFTALNGTNGSVEADGLV 300
DB 241 GALIQSVKCLADVAVLTTFCLSVFALIGLOFMGNLHKCVRNFTALNGTNGSVEADGLV 300
OY 301 WESLDLYSDPENYLNKGTSDVLLCGNSSDAGTCPEGRCLKXGENPDHGYTSPDSFAW 360
DB 301 WESLDLYSDPENYLNKGTSDVLLCGNSSDAGTCPEGRCLKXGENPDHGYTSPDSFAW 360
OY 361 AFLALPFLMQDQCEBRLYQOTLRSAKIYIMFFMLVIFLGSFVLNLIIVAVAYEON 420
DB 361 AFLALPFLMQDQCEBRLYQOTLRSAKIYIMFFMLVIFLGSFVLNLIIVAVAYEON 420
OY 421 QATIAETEEKRFRQEMEMLKKEHALITRGVDVTRSSSIENSPLAPVNSHERSKRRK 480
DB 421 QATIAETEEKRFRQEMEMLKKEHALITRGVDVTRSSSIENSPLAPVNSHERSKRRK 480
OY 481 RMSSGTECCGDRLPKDSDEGPRAMNHLSTRGLSTSMKPRSSRSISFFRRRDGSE 540
DB 481 RMSSGTECCGDRLPKDSDEGPRAMNHLSTRGLSTSMKPRSSRSISFFRRRDGSE 540
OY 541 ADPADDENSTAGESESHRTSLVPMPLRRTSAGQSPGTSAFGHALHGKKNSTVDCNGV 600
DB 541 ADPADDENSTAGESESHRTSLVPMPLRRTSAGQSPGTSAFGHALHGKKNSTVDCNGV 600
OY 601 VSLGAGDPKATSPGSHILARPVMLEHPDPTTSPSEEGPQMLTSGAPCVDGFEFGARQ 660
DB 601 VSLGAGDPKATSPGSHILARPVMLEHPDPTTSPSEEGPQMLTSGAPCVDGFEFGARQ 660
OY 661 PALNAVSVLTGALTELESRHKCPKCNWRLAQRVLIMECCGLMMSIKQGVLYVMDPPTD 720
DB 661 PALNAVSVLTGALTELESRHKCPKCNWRLAQRVLIMECCGLMMSIKQGVLYVMDPPTD 720
OY 721 LITTMCIYVLTLPALTEHYNTSEFEEMLVGNVLTGTFPAEMTFKIALDPYYPQOG 780
DB 721 LITTMCIYVLTLPALTEHYNTSEFEEMLVGNVLTGTFPAEMTFKIALDPYYPQOG 780
OY 781 WNIPEDSIVILSLMELGSRMSNTSVLRSPFLRVFKLAKSWPLNTLIKIGNSVGLG 840
DB 781 WNIPEDSIVILSLMELGSRMSNTSVLRSPFLRVFKLAKSWPLNTLIKIGNSVGLG 840


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QY 841 NLTLLVIAIVFIFAVVGMQLFGKNTYSELSDSDGLPRHMMDFPHAFILIRILCGEMI 900
DB 841 NLTLLVIAIVFIFAVVGMQLFGKNTYSELSDSDGLPRHMMDFPHAFILIRILCGEMI 900
QY 901 ETMDMEVSGSLCLVFLVWVIGNLVNLFTALLSPSADNLTADEDEMMNLQ 960
DB 901 ETMDMEVSGSLCLVFLVWVIGNLVNLFTALLSPSADNLTADEDEMMNLQ 960
QY 961 LALANIORGLARVKTITMDPCCGLLRORPORPALAAGOLPSCIATPYSPPEETKXP 1020
DB 961 LALANIORGLARVKTITMDPCCGLLRORPORPALAAGOLPSCIATPYSPPEETKXP 1020
QY 1021 PPRKTRFEEGBEGPOGTPGDEPEYCVPIAVASEPTDDEBDEENSLGTEEESSKQESOP 1080
DB 1021 PPRKTRFEEGBEGPOGTPGDEPEYCVPIAVASEPTDDEBDEENSLGTEEESSKQESOP 1080
QY 1081 VSGGEAPPPSRKTRWQVATASSEASASQADMRQWKAEPQACGETPEPSCSEGST 1140
DB 1081 VSGGEAPPPSRKTRWQVATASSEASASQADMRQWKAEPQACGETPEPSCSEGST 1140
QY 1141 ADMNTAIELBQIPDLGQDVKDPEDCFTBGCVRRCPCCAVDTTQAPGKVMRLRKTCTHI 1200
DB 1141 ADMNTAIELBQIPDLGQDVKDPEDCFTBGCVRRCPCCAVDTTQAPGKVMRLRKTCTHI 1200
QY 1201 VEHSMFETPIIFMILLSSGALAFEDILYERKTIKYLEVADKMTYVVLKMLKWAY 1260
DB 1201 VEHSMFETPIIFMILLSSGALAFEDILYERKTIKYLEVADKMTYVVLKMLKWAY 1260
QY 1261 GPKKFTNAMCWLDELVDVSLVSVANTLGFENGPILKSLRTLRALPLRALSRFEGMR 1320
DB 1261 GPKKFTNAMCWLDELVDVSLVSVANTLGFENGPILKSLRTLRALPLRALSRFEGMR 1320
QY 1321 VVNALVGAIPBIMAVLLVCLIFMLIFSIMGNLFAKRGRCINOTEGDLPLNTYIVNNK 1380
DB 1321 VVNALVGAIPBIMAVLLVCLIFMLIFSIMGNLFAKRGRCINOTEGDLPLNTYIVNNK 1380
QY 1381 SOCESLNTLGEIYMKVKNPENNVAGYIALIQVATPFKGMIDIMAAVDSGYEERQOME 1440
DB 1381 SOCESLNTLGEIYMKVKNPENNVAGYIALIQVATPFKGMIDIMAAVDSGYEERQOME 1440
QY 1441 YNLVYIYFVIFIIIFGSEFTLNLFIGVLIIDNNOQKKLGGODIPMTEBOKKYANMKL 1500
DB 1441 YNLVYIYFVIFIIIFGSEFTLNLFIGVLIIDNNOQKKLGGODIPMTEBOKKYANMKL 1500
QY 1501 GSKKQKPIPRPLNKYGFIDIVTKQAPDVTIMELCLINVTMMVETDQSPKINILLA 1560
DB 1501 GSKKQKPIPRPLNKYGFIDIVTKQAPDVTIMELCLINVTMMVETDQSPKINILLA 1560
QY 1561 KINLIFVAFIPTGECIVKLAALRHYYFTNSMNFEDVVVILSVGVVLSDIIOKXFFSPTL 1620
DB 1561 KINLIFVAFIPTGECIVKLAALRHYYFTNSMNFEDVVVILSVGVVLSDIIOKXFFSPTL 1620
QY 1621 FRVIRLARIIGRLIRIRAGAKGIRTLPLMMSLPALFINGILLFLVMFYISIFGMANAY 1680
DB 1621 FRVIRLARIIGRLIRIRAGAKGIRTLPLMMSLPALFINGILLFLVMFYISIFGMANAY 1680
QY 1681 VKMEAGIDDMFNPOTFANSMLCLFOITTSAGMDGLSLPILNTGPPYCDPTLPNSNGSGD 1740
DB 1681 VKMEAGIDDMFNPOTFANSMLCLFOITTSAGMDGLSLPILNTGPPYCDPTLPNSNGSGD 1740
QY 1741 CGSPAVGLLFTTYYIIISFLIVVMNYIAIILENPSVAIBESBEPISBDPFMYITWKF 1800
DB 1741 CGSPAVGLLFTTYYIIISFLIVVMNYIAIILENPSVAIBESBEPISBDPFMYITWKF 1800
QY 1801 DPEATQIFIEYVLSDFADALSPLRIAKPNOISLNMOLPMVSGDRHICMOLFAFTGRV 1860
DB 1801 DPEATQIFIEYVLSDFADALSPLRIAKPNOISLNMOLPMVSGDRHICMOLFAFTGRV 1860
QY 1861 LGESGEMDALKIOMEKTMANPSKISTEPIITTLRRRHEVSANVIOQAFRRHLQSL 1920
DB 1861 LGESGEMDALKIOMEKTMANPSKISTEPIITTLRRRHEVSANVIOQAFRRHLQSL 1920
QY 1921 KHASLFRQOAGSGISEEDAPBRGLIYVMSSENSRRLGPPSSSSISSTSPPEYDSVT 1980

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DB 1921 KHASLFRQOAGSGISEEDAPBRGLIYVMSSENSRRLGPPSSSSISSTSPPEYDSVT 1980
QY 1981 RATSNDLQVRGSDYSHSEDLADFPSPDRDRESIV 2015
DB 1981 RATSNDLQVRGSDYSHSEDLADFPSPDRDRESIV 2015

RESULT 11
AAB82239
ID AAB82239 standard; protein; 2016 AA.
XX
AC AAB82239;
XX
DT 21-JUN-2001 (first entry)
XX
DE Human SCNSA protein.
XX
KW SCNSA; Long QT syndrome; LQTS; cardiovascular disease;
XX Romano-Ward syndrome; diagnosis; prognosis; therapy; drug screening.
XX
OS Homo sapiens.
XX
PN MO200124681-A2.
XX
PD 12-APR-2001.
XX
PF 09-AUG-2000; 2000WO-US021660.
XX
PR 09-AUG-1999; 99US-0147488P.
XX
PR 17-MAR-2000; 2000US-0190057P.
XX
PA (UTAH ) UNIV UTAH RES FOUND.
XX
PI Keating MT, Splawski I;
XX
DR MPI; 2001-290564/30.
XX
DR N-PSDB; AAF30825.
XX
PT New KVLQT1 and SCNSA genes, which contains alterations or mutations,
XX useful in diagnostic/prognostic or drug screening methods, particularly in
XX mutational analyses for screening individuals with or at risk for long QT
XX syndrome.
XX
PS Claim 31; Page 69-75; 76pp; English.
XX
CC The present sequence is that of the protein encoded by the human SCNSA
XX gene. This gene is implicated in Romano-Ward syndrome, the autosomal
XX dominant form of Long QT syndrome (LQTS). Novel mutations have been
XX identified in the gene using single strand conformation polymorphism
XX analysis. These result in the following amino acid alterations: D114N,
XX L1501V, del1617, R1623L, E1784K and S1787N. Isolated human polypeptides
XX comprising such a mutation (see AAB82240-45) are claimed. Knowledge of
XX the mutations provides means for assessing a risk in a human subject for
XX LQTS, for diagnosing a mutation which causes LQTS, and for screening for
XX drugs useful in treating a human having a mutation in the SCNSA gene
XX
SQ Sequence 2016 AA;
XX
Query Match 99.5%; Score 10432.5; DB 4; Length 2016;
Best Local Similarity 99.5%; Pred. No. 0;
Matches 2006; Conservative 2; Mismatches 7; Indels 1; Gaps 1;
QY 1 MANFLPRGTSSFRFTRESLAIEKMAEKQAGSTTLQSRGGLPEEAPRPQDLQQA 60
DB 1 MANFLPRGTSSFRFTRESLAIEKMAEKQAGSTTLQSRGGLPEEAPRPQDLQQA 60
QY 61 SKKLPDIYGNPQOLISEPLLEDLPFYSTQKTFIVANKKGTIFPFSATNALYVSPRPPI 120
DB 61 SKKLPDIYGNPQOLISEPLLEDLPFYSTQKTFIVANKKGTIFPFSATNALYVSPRPPI 120
QY 121*GRAAVKILVSLFNLIMCTLITNCVMAQHDPPMTKYVEYPTATYTFESLVKILARG 180

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Db 121 RRAAVKILVHSLFNMILMCTILITNVCVMAQHDPPWTKYVEYTFALTYTESLYKILARA 180
 QY 181 FCLHAFPLRDPWMWMLDFSVIIMAYTTEFVDLGNVSALRFRVLRALKTISVIGLKTIV 240
 Db 181 FCLHAFPLRDPWMWMLDFSVIIMAYTTEFVDLGNVSALRFRVLRALKTISVIGLKTIV 240
 QY 241 GALLQSVKCLADVWVLTWPCISVFPALIGLOLPMNGLRHKCVRNPTALNGTNGSVEADOLV 300
 Db 241 GALLQSVKCLADVWVLTWPCISVFPALIGLOLPMNGLRHKCVRNPTALNGTNGSVEADOLV 300
 QY 301 MESIDLYSDPENYLLKNGSTDVLLCGNSSDAGTCEPEGYRCLAKGENPDHGYTFSDFRW 360
 Db 301 MESIDLYSDPENYLLKNGSTDVLLCGNSSDAGTCEPEGYRCLAKGENPDHGYTFSDFRW 360
 QY 361 APLALFRLMTQDCWERYLQOTLRSAKGIYMIFFMLVIFLGSPPYLVNLLAVAVAMAYEON 420
 Db 361 APLALFRLMTQDCWERYLQOTLRSAKGIYMIFFMLVIFLGSPPYLVNLLAVAVAMAYEON 420
 QY 421 QATTAEETEEKRFRPEAMEMLKKEHEALTIRGVTVSSSLSEMSPLAVNSHERSKRRK 480
 Db 421 QATTAEETEEKRFRPEAMEMLKKEHEALTIRGVTVSSSLSEMSPLAVNSHERSKRRK 480
 QY 481 RMSSTGECGEDRLPKSDSEDPGRAMNHLSTRGLSRTSMKPRSRSRGSIFTFRRDLGSE 540
 Db 481 RMSSTGECGEDRLPKSDSEDPGRAMNHLSTRGLSRTSMKPRSRSRGSIFTFRRDLGSE 540
 QY 541 ADFADENSTAGESESHRTSLILVWPLRRTSAQGGPSGTSAPGHALHKKONSTDVNCV 600
 Db 541 ADFADENSTAGESESHRTSLILVWPLRRTSAQGGPSGTSAPGHALHKKONSTDVNCV 600
 QY 601 VSLGAGPPEARTSPSSHLLRPVMLEHPDPTTSPSEPPGQMLTSSQAQCVGFEERGRQ 660
 Db 601 VSLGAGPPEARTSPSSHLLRPVMLEHPDPTTSPSEPPGQMLTSSQAQCVGFEERGRQ 660
 QY 661 RALSAYSVLTSALEELIESRHKPCPCMNRLAQRVYIWECCPLMSIKOGVXLVWMDPPTD 720
 Db 661 RALSAYSVLTSALEELIESRHKPCPCMNRLAQRVYIWECCPLMSIKOGVXLVWMDPPTD 720
 QY 721 LTTTNCIVLNTLFMALIEHNMSTSEFEEMLOVGNLVTGIFTAEMTFKIIALDPYYFQOG 780
 Db 721 LTTTNCIVLNTLFMALIEHNMSTSEFEEMLOVGNLVTGIFTAEMTFKIIALDPYYFQOG 780
 QY 781 WNIPISTIIVILSMLGLSRMSNLVLSRFLLRVFKLAKSWPTLNTLIIKIIGNSVGLG 840
 Db 781 WNIPISTIIVILSMLGLSRMSNLVLSRFLLRVFKLAKSWPTLNTLIIKIIGNSVGLG 840
 QY 841 NLTLVLAIVFIFAVVGMQLPGKNYSELSDSDGLPRWMDPFHAFILIFRILCGEMI 900
 Db 841 NLTLVLAIVFIFAVVGMQLPGKNYSELSDSDGLPRWMDPFHAFILIFRILCGEMI 900
 QY 901 ETTMDCMEVSGSLCLVFLVAVIIGNLVNLFLALLISFSADNLTAPDEDEMANILO 960
 Db 901 ETTMDCMEVSGSLCLVFLVAVIIGNLVNLFLALLISFSADNLTAPDEDEMANILO 960
 QY 961 LALATIORGLRFRVKTTPMFCGGLRORPKPAAALAAQOLRSCATPSPPEETERYP 1020
 Db 961 LALATIORGLRFRVKTTPMFCGGLRORPKPAAALAAQOLRSCATPSPPEETERYP 1020
 QY 1021 PTRKETRFEGEGOPGGTGPDEPEVCVPIAVAESDTPDOEBDEENSLGTEESSK-OESQ 1079
 Db 1021 PTRKETRFEGEGOPGGTGPDEPEVCVPIAVAESDTPDOEBDEENSLGTEESSK-OESQ 1080
 QY 1080 PVSQGEADPDSRTMSQVSATASSEAEASASQADWQOMKAPQAPGCGETPDSCEGS 1139
 Db 1081 PVSQGEADPDSRTMSQVSATASSEAEASASQADWQOMKAPQAPGCGETPDSCEGS 1140
 QY 1140 TADMNTTAALEBQIPRLGQVDPEDCFEGECYRRPCCAVDTTQAPGVWMLRKTCTH 1199
 Db 1141 TADMNTTAALEBQIPRLGQVDPEDCFEGECYRRPCCAVDTTQAPGVWMLRKTCTH 1200
 QY 1200 IYEHSMFEFTIIFMILLSSGALAFEDIVLEBRKTIIVLLEVDKMPYTYFVLEMLLKNVA 1259
 Db 1201 IYEHSMFEFTIIFMILLSSGALAFEDIVLEBRKTIIVLLEVDKMPYTYFVLEMLLKNVA 1260

QY 1260 YGFRKYFTNMACWLDPLIVDVSLVSVANTLGAEMGPISKSLRTLRLALRPLASPEGM 1319
 Db 1261 YGFRKYFTNMACWLDPLIVDVSLVSVANTLGAEMGPISKSLRTLRLALRPLASPEGM 1320
 QY 1320 RYVYNALVGAIPSIMVNLVCLIFWLIFSIMGVNLPAKFGRCINOTEGDPLNYTTVNN 1379
 Db 1321 RYVYNALVGAIPSIMVNLVCLIFWLIFSIMGVNLPAKFGRCINOTEGDPLNYTTVNN 1380
 QY 1380 KSQCESLNTGELWTKVKNFNVGAGYALLOVATPKKMNIMTAAVUSRCYEEQPOW 1439
 Db 1381 KSQCESLNTGELWTKVKNFNVGAGYALLOVATPKKMNIMTAAVUSRCYEEQPOW 1440
 QY 1440 EYMLYMYTVFVIFPIESGFETLNLFGVILIDNFNOQKKUGGODIPMTTEOKKYNNAMK 1499
 Db 1441 EYMLYMYTVFVIFPIESGFETLNLFGVILIDNFNOQKKUGGODIPMTTEOKKYNNAMK 1500
 QY 1500 LGSKKPQKPIPRPLNKYQGFIPDIVTKQADVTIMFLICLNNYTMVETDQSPKINIL 1559
 Db 1501 LGSKKPQKPIPRPLNKYQGFIPDIVTKQADVTIMFLICLNNYTMVETDQSPKINIL 1560
 QY 1560 AKINLFLVAIFTEGCIVKLAALRHYFTNSWNIFFDFVVLVLSVGYTLSDIIOKYFSP 1619
 Db 1561 AKINLFLVAIFTEGCIVKLAALRHYFTNSWNIFFDFVVLVLSVGYTLSDIIOKYFSP 1620
 QY 1620 LFRVIRIARIGRIILRLRGAKGIRTLFLPALMGLPALFNIGLLFLVMFIYSIFGMANFA 1679
 Db 1621 LFRVIRIARIGRIILRLRGAKGIRTLFLPALMGLPALFNIGLLFLVMFIYSIFGMANFA 1680
 QY 1680 YVKWEAGIDMFMFOFPANSMLCLFOITTSAGMDGLSPILANTGPPYCDPTLPNSNGSRG 1739
 Db 1681 YVKWEAGIDMFMFOFPANSMLCLFOITTSAGMDGLSPILANTGPPYCDPTLPNSNGSRG 1740
 QY 1740 DCGSPAVGILFTTYYIIISFLIVYNNKIAIILLENFSVATEESTEPLESDPFMEYETWEK 1799
 Db 1741 DCGSPAVGILFTTYYIIISFLIVYNNKIAIILLENFSVATEESTEPLESDPFMEYETWEK 1800
 QY 1800 FDPBATOFIEVSVDPADLSEPLRAXPNQISLIMNDLPMYSGDRHICMDILFAFTRK 1859
 Db 1801 FDPBATOFIEVSVDPADLSEPLRAXPNQISLIMNDLPMYSGDRHICMDILFAFTRK 1860
 QY 1860 VLGSSEMDALKIOEEKFMAANPSKISYEBITTTLRKHEEVSAMVIOQAFRRHLLORS 1919
 Db 1861 VLGSSEMDALKIOEEKFMAANPSKISYEBITTTLRKHEEVSAMVIOQAFRRHLLORS 1920
 QY 1920 LKHASFLPROQAGSGLSEEDAPEREGLIAYVMSNERSPLCPBSSSISSTSPSPSYDSV 1979
 Db 1921 LKHASFLPROQAGSGLSEEDAPEREGLIAYVMSNERSPLCPBSSSISSTSPSPSYDSV 1980
 QY 1980 TRATSNDLQVNGSDYSHSEDLADPPSPDRDRESIV 2015
 Db 1981 TRATSNDLQVNGSDYSHSEDLADPPSPDRDRESIV 2016

RESULT 12 4
 ADD44756
 ID ADD44756 standard; protein; 2016 AA.
 XX
 AC ADD44756;
 DT 29-JAN-2004 (first entry)
 XX
 DE Human Protein Q14524, SEQ ID NO 10185.
 XX
 KW Human; pain; neuronal tissue; gene therapy;
 KW spinal segmental nerve injury; chronic constriction injury; CCI;
 KW spared nerve injury; SNJ; Chung.
 OS Homo sapiens.
 PN WO2003016475-A2.
 XX
 PD 27-FEB-2003.

XX 14-AUG-2002; 2002MO-US025765.
PF
XX 14-AUG-2001; 2001US-0312147P.
PR 01-NOV-2001; 2001US-0346382P.
PR 26-NOV-2001; 2001US-0333347P.
XX
XX (GHEO) GEN HOSPITAL CORP.
PA (FARB) BAYER AG.
XX
PI Woolf C, D'urso D, Befort K, Costigan M;
XX WPI; 2003-268312/26.
DR GENBANK; Q14524.
XX
XX
PT New composition comprising two or more isolated polypeptides, useful for
PT preparing a medicament for treating pain in an animal.
PS
XX Claim 1; Page: 1017pp; English.
XX
XX The invention discloses a composition comprising two or more isolated rat
CC or human polynucleotides or a polynucleotide which represents a fragment,
CC derivative or allelic variation of the nucleic acid sequence. Also
CC claimed are a vector comprising the novel polynucleotide, a host cell
CC comprising the vector, a method for identifying a nucleotide sequence
CC which is differentially regulated in an animal subjected to pain and a
CC kit to perform the method, an array, a method for identifying an agent
CC that increases or decreases the expression of the polynucleotide sequence
CC that is differentially expressed in neuronal tissue of a first animal
CC subjected to pain, a method for identifying a compound which regulates
CC the expression of a polynucleotide sequence which is differentially
CC expressed in an animal subjected to pain, a method for identifying a
CC compound that regulates the activity of one or more of the
CC polynucleotides, a method for producing a pharmaceutical composition, a
CC method for identifying a compound or small molecule that regulates the
CC activity of one or more of the polypeptides given in the
CC specification, a method for identifying a compound useful in treating
CC pain and a pharmaceutical composition comprising the one or more
CC polypeptides or their antibodies. The polynucleotide or the compound that
CC modulates its activity is useful for preparing a medicament for treating
CC pain (e.g. spinal segmental nerve injury (Chung), chronic constriction
CC injury (CCI) and spared nerve injury (SNI)) in an animal (e.g. gene
CC therapy). The sequence presented is a human protein (shown in Table 2 of
CC the specification) which is differentially expressed during pain. Note:
CC The sequence data for this patent did not form part of the printed
CC specification, but was obtained in electronic form directly from WIPO at
CC ftp.wipo.int/pub/published_pct_sequences.
XX
XX Sequence 2016 AA:
SQ

Query Match 99.5%; Score 10432.5; DB 7; Length 2016;
Best Local Similarity 99.5%; Pred. No. 0;
Matches 2006; Conservative 2; Mismatches 7; Indels 1; Gaps 1;

QY 1 MANPLPGTSGFRFTRESLSAIEKMAEKQARSTLQBSRELPBEARPOLDIOA 60
DB 1 MANPLPGTSGFRFTRESLSAIEKMAEKQARSTLQBSRELPBEARPOLDIOA 60
QY 61 SKKLPLDLYGNPQOELIGEPLEBDLPFYSTOKTFIYLNKQKTLFRFSATNALVYLSPPHPI 120
DB 61 SKKLPLDLYGNPQOELIGEPLEBDLPFYSTOKTFIYLNKQKTLFRFSATNALVYLSPPHPI 120
QY 121 RRAAVKIIIVHSLENNLIMCTIITNCVFMAQNDPPWTKYVEYTFPAITPESLVKILARG 180
DB 121 RRAAVKIIIVHSLENNLIMCTIITNCVFMAQNDPPWTKYVEYTFPAITPESLVKILARG 180
QY 181 FCLHAFETLRDPMNMLDSVIIIMATYTERVUDIGNVSALRTFRVYALAKTIVISLKTIV 240
DB 181 FCLHAFETLRDPMNMLDSVIIIMATYTERVUDIGNVSALRTFRVYALAKTIVISLKTIV 240
QY 241 GALIOSVKKLADVMVLTFCISVFALIGLQIFMGNIIRHKCVANFTALNGTNGSVEADGLV 300
DB 241 GALIOSVKKLADVMVLTFCISVFALIGLQIFMGNIIRHKCVANFTALNGTNGSVEADGLV 300

QY 301 WESLDLYSDPENYLLKNGTSDVLLCGNSSDAGTCEGYRCLKAGENPDHGYTSFDSFAM 360
DB 301 WESLDLYSDPENYLLKNGTSDVLLCGNSSDAGTCEGYRCLKAGENPDHGYTSFDSFAM 360
QY 361 AFLALFRIMTODCWERLYOQTLRSAGKIYMFVMLVIFLGSFYLVNLLAVVAMAYEBQN 420
DB 361 AFLALFRIMTODCWERLYOQTLRSAGKIYMFVMLVIFLGSFYLVNLLAVVAMAYEBQN 420
QY 421 QATIAETEKEKRFQEAEMEMKKEHEALTITIGVTVSSLSLEMSPLAVNHSERSKRRK 480
DB 421 QATIAETEKEKRFQEAEMEMKKEHEALTITIGVTVSSLSLEMSPLAVNHSERSKRRK 480
QY 481 RMSGTECGDRLPKPSDEGPPRAMNLTSTRGLSRTSMKPRSRGSIFFRRRDLGSE 540
DB 481 RMSGTECGDRLPKPSDEGPPRAMNLTSTRGLSRTSMKPRSRGSIFFRRRDLGSE 540
QY 541 ADPADDENSTAGESESHRTSLVWPFLRTSAQGPSPGTSAPGHALHGXKXSTYDCNGV 600
DB 541 ADPADDENSTAGESESHRTSLVWPFLRTSAQGPSPGTSAPGHALHGXKXSTYDCNGV 600
QY 601 VSLGAGDPEATSPGSHLLRPVMLEHPDPTTSPSEPGQOMLTSQAPCVDFEERPARQ 660
DB 601 VSLGAGDPEATSPGSHLLRPVMLEHPDPTTSPSEPGQOMLTSQAPCVDFEERPARQ 660
QY 661 RALSNVSVLTSALELESERHKPCPCNRLAORVLIWCCCLUMSIRKGVULVWMDPFD 720
DB 661 RALSNVSVLTSALELESERHKPCPCNRLAORVLIWCCCLUMSIRKGVULVWMDPFD 720
QY 721 LTIMCIYANTLPMALHEYNMTSEFEEMLOVGNLFTGSIPTAEMTFKIALDPYYPFOG 780
DB 721 LTIMCIYANTLPMALHEYNMTSEFEEMLOVGNLFTGSIPTAEMTFKIALDPYYPFOG 780
QY 781 WNIPDSIIIVILSLMEILGSRMSNTSVRSFRLIRVFKLAKSWPTLNTLKIIGNSVGLG 840
DB 781 WNIPDSIIIVILSLMEILGSRMSNTSVRSFRLIRVFKLAKSWPTLNTLKIIGNSVGLG 840
QY 841 NLTVLAIIVIFAVVGQMLRGKXYSERLSDSGILPWHMMDPFAHLIIFRIICGEMI 900
DB 841 NLTVLAIIVIFAVVGQMLRGKXYSERLSDSGILPWHMMDPFAHLIIFRIICGEMI 900
QY 901 ETTMDCHEVSGOSCLIVFLVWVIGNLVNLFALILSLSSFSADNLTAPEDEBEMNLQ 960
DB 901 ETTMDCHEVSGOSCLIVFLVWVIGNLVNLFALILSLSSFSADNLTAPEDEBEMNLQ 960
QY 961 LALARIQGLRFVAKRTIWDPCGILLRORPQKPAALAAQGLPSCIATPYSPPETEXVP 1020
DB 961 LALARIQGLRFVAKRTIWDPCGILLRORPQKPAALAAQGLPSCIATPYSPPETEXVP 1020
QY 1021 PTRKETRPEEBEGOGCTPGDPPEVCVPIVAVESPTDDOEEDENSIGTEBESSR-QSSQ 1079
DB 1021 PTRKETRPEEBEGOGCTPGDPPEVCVPIVAVESPTDDOEEDENSIGTEBESSR-QSSQ 1079
QY 1080 PVSGGPEAPPDPSRTWSQVATASSEABASAOADWROOKABPOAPGCGTPEBDSGSG 1139
DB 1080 PVSGGPEAPPDPSRTWSQVATASSEABASAOADWROOKABPOAPGCGTPEBDSGSG 1139
QY 1140 TADMNTNALLBOJIPDLGQDVXDPEDCFTBGCYVRCPCCAVDTTQAPGKWMRLKTCYH 1199
DB 1140 TADMNTNALLBOJIPDLGQDVXDPEDCFTBGCYVRCPCCAVDTTQAPGKWMRLKTCYH 1199
QY 1200 IVHSWPEFTFIIFMILSLSSGALAFBDIYLERKTIKYULEYADKMFYVVFLEMLLKVA 1259
DB 1200 IVHSWPEFTFIIFMILSLSSGALAFBDIYLERKTIKYULEYADKMFYVVFLEMLLKVA 1259
QY 1260 YGFKKYFTNACMIDFLIVDSVLSVANTGAPAEKGIKSLRTLRALRPLRALSREFGM 1319
DB 1260 YGFKKYFTNACMIDFLIVDSVLSVANTGAPAEKGIKSLRTLRALRPLRALSREFGM 1319
QY 1320 RVVYNALVGALPSIMNVLLVCLIFWLIFSLINGVLPACKFGRCINQTEGDLPLNVTYVNN 1379
DB 1320 RVVYNALVGALPSIMNVLLVCLIFWLIFSLINGVLPACKFGRCINQTEGDLPLNVTYVNN 1379
QY 1321 RVVYNALVGALPSIMNVLLVCLIFWLIFSLINGVLPACKFGRCINQTEGDLPLNVTYVNN 1380
DB 1321 RVVYNALVGALPSIMNVLLVCLIFWLIFSLINGVLPACKFGRCINQTEGDLPLNVTYVNN 1380

QY 1380 KSCCESLNLGELIWTYKRVNPNVAGYIALLLQVATPKGMDIMYAAVDSRGYEQPW 1439
 DB 1381 KSCCESLNLGELIWTYKRVNPNVAGYIALLLQVATPKGMDIMYAAVDSRGYEQPW 1440
 QY 1440 EYNLYMYTYPIYFIIFGSPFTLNLPFIYIINDFNQKKKGGGODIEMTEBOKKYYNMMK 1499
 DB 1441 EYNLYMYTYPIYFIIFGSPFTLNLPFIYIINDFNQKKKGGGODIEMTEBOKKYYNMMK 1500
 QY 1500 LGSKKPKQPIPRPLNKYQGFIPDIYTKQAFDVTIWFELIINMVTMTVETDQSEKINIL 1559
 DB 1501 LGSKKPKQPIPRPLNKYQGFIPDIYTKQAFDVTIWFELIINMVTMTVETDQSEKINIL 1560
 QY 1560 AKINILFPAITFGECIVLALAPHYFNNSNNIPFVVVILISYGVLSIDIIQKYPSPPT 1619
 DB 1561 AKINILFPAITFGECIVLALAPHYFNNSNNIPFVVVILISYGVLSIDIIQKYPSPPT 1620
 QY 1620 LFRVRLARIGRLILKIGAKGIRTLPALMMSLPALNIGLLFLVMFIYSFGMANFA 1679
 DB 1621 LFRVRLARIGRLILKIGAKGIRTLPALMMSLPALNIGLLFLVMFIYSFGMANFA 1680
 QY 1680 YVKNWAGIDDMENFQTFPANSMLCLFOITTSAGMDGLSPIINTGPPYCDPTLPNSNGSRG 1739
 DB 1681 YVKNWAGIDDMENFQTFPANSMLCLFOITTSAGMDGLSPIINTGPPYCDPTLPNSNGSRG 1740
 QY 1740 DCGSPANGILPFTTYIIISPLIVNMXYAIIENFSVATPESSTPELSEDDPMFEIEMK 1799
 DB 1741 DCGSPANGILPFTTYIIISPLIVNMXYAIIENFSVATPESSTPELSEDDPMFEIEMK 1800
 QY 1800 FDPPEATQFIEYSVLSDPADALSEPRLAKENQISLINDLPMVSGDRHICMDILPAFTKR 1859
 DB 1801 FDPPEATQFIEYSVLSDPADALSEPRLAKENQISLINDLPMVSGDRHICMDILPAFTKR 1860
 QY 1860 VLGSSEGDMDALKIOMEKFKMANPSKISYEPIITTLRKHEEVSAMVIOAFRRHLLORS 1919
 DB 1861 VLGSSEGDMDALKIOMEKFKMANPSKISYEPIITTLRKHEEVSAMVIOAFRRHLLORS 1920
 QY 1920 LKHAFLFRQOAGSGISEEDAPERGLAYVMSNPSPRLGPPSSSISSTSPSPSYDSV 1979
 DB 1921 LKHAFLFRQOAGSGISEEDAPERGLAYVMSNPSPRLGPPSSSISSTSPSPSYDSV 1980
 QY 1980 TRATSDNLQVRGSDYSHSEDLADFPSPSPDRRESIV 2015
 DB 1981 TRATSDNLQVRGSDYSHSEDLADFPSPSPDRRESIV 2016

RESULT 13

ADE55106
ID ADE55106 standard; protein; 2016 AA.

XX ADE55106;

DT 29-JAN-2004 (first entry)

DE Human Protein NP_000326, SEQ ID NO 911.

XX Human; pain; neuronal tissue; gene therapy;
KW spinal segmental nerve injury; chronic constriction injury; CCI;
KW spared nerve injury; SNI; Chung.

XX Homo sapiens.

OS WO0203016475-A2.

PN 27-FEB-2003.

PD 14-AUG-2002; 2002WO-US025765.

PF 14-AUG-2001; 2001US-0312147P.

PR 01-NOV-2001; 2001US-0346382P.

PR 26-NOV-2001; 2001US-033347P.

XX (GEHO) GEN HOSPITAL CORP.

PA (FARB) BAYER AG.

XX Woolf C, D'urso D, Befort K, Coatsigan M;
 PI WPI; 2003-268312/26.
 XX GENBANK; NP_000326.
 DR
 DR
 PT New composition comprising two or more isolated polypeptides, useful for
 PT preparing a medicament for treating pain in an animal.
 PS Claim 1, Page; 1017p; English.

CC The invention discloses a composition comprising two or more isolated rat
 CC or human polynucleotides or a polynucleotide which represents a fragment,
 CC derivative or allelic variation of the nucleic acid sequence. Also
 CC comprising the vector, a method for identifying a nucleotide sequence
 CC which is differentially regulated in an animal subjected to pain and a
 CC kit to perform the method, an array, a method for identifying an agent
 CC that increases or decreases the expression of the polynucleotide sequence
 CC that is differentially expressed in neuronal tissue of a first animal
 CC subjected to pain, a method for identifying a compound which regulates
 CC the expression of a polynucleotide sequence which is differentially
 CC expressed in an animal subjected to pain, a method for identifying a
 CC compound that regulates the activity of one or more of the
 CC polynucleotides, a method for producing a pharmaceutical composition, a
 CC method for identifying one or more of the polypeptides given in the
 CC activity in an animal of one or more of the polypeptides useful in treating
 CC specification, a method for identifying a compound useful in treating
 CC pain and a pharmaceutical composition comprising the one or more
 CC polypeptides or their antibodies. The polynucleotide or the compound that
 CC modulates its activity is useful for preparing a medicament for treating
 CC pain (e.g. spinal segmental nerve injury (SNI), chronic constriction
 CC injury (CCI) and spared nerve injury (SNI) in an animal (e.g. gene
 CC therapy). The sequence presented is a human protein (shown in Table 2 of
 CC the specification) which is differentially expressed during pain. Note:
 CC The sequence data for this patent did not form part of the printed
 CC specification, but was obtained in electronic form directly from WIPO at
 CC ftp.wipo.int/pub/published_pcl_sequences.

SQ Sequence 2016 AA;

Query Match 99.5%; Score 10432.5; DB 7; Length 2016;
 Best Local Similarity 99.5%; Pred. No. 0;
 Matches 2006; Conservative 2; Mismatches 7; Indels 1; Gaps 1;

QY 1 MANFLPRGTSSEFRFRRESLAIEKMAEKQAGSTTLQESRGLPEEAPRQQLQA 60
 DB 1 MANFLPRGTSSEFRFRRESLAIEKMAEKQAGSTTLQESRGLPEEAPRQQLQA 60
 QY 61 SKKLPLDYGNPQELIGPELEDLPFYSTOKTFIVLNKKGTIPFSAATNALYVLSPPHPI 120
 DB 61 SKKLPLDYGNPQELIGPELEDLPFYSTOKTFIVLNKKGTIPFSAATNALYVLSPPHPI 120
 QY 121 RRAAVKILVHSLFNNLIMCTILNVCVMAQDHPDPMTKYVYETPTAIYTESGLYKILARG 180
 DB 121 RRAAVKILVHSLFNNLIMCTILNVCVMAQDHPDPMTKYVYETPTAIYTESGLYKILARG 180
 QY 121 RRAAVKILVHSLFNNLIMCTILNVCVMAQDHPDPMTKYVYETPTAIYTESGLYKILARG 180
 DB 121 RRAAVKILVHSLFNNLIMCTILNVCVMAQDHPDPMTKYVYETPTAIYTESGLYKILARG 180
 QY 181 FCLHAFTFLRPPNWDLPDSVIMAYTTEFVULGNVSLARTFRVRLAKTISVIGSLKTIY 240
 DB 181 FCLHAFTFLRPPNWDLPDSVIMAYTTEFVULGNVSLARTFRVRLAKTISVIGSLKTIY 240
 QY 181 FCLHAFTFLRPPNWDLPDSVIMAYTTEFVULGNVSLARTFRVRLAKTISVIGSLKTIY 240
 DB 181 FCLHAFTFLRPPNWDLPDSVIMAYTTEFVULGNVSLARTFRVRLAKTISVIGSLKTIY 240
 QY 241 GALIISVKKGLADVWVLFVFCLSVPALIGLQLEFNGNLBHKCVRNFTALNGTNGSYEADGLV 300
 DB 241 GALIISVKKGLADVWVLFVFCLSVPALIGLQLEFNGNLBHKCVRNFTALNGTNGSYEADGLV 300
 QY 301 WESIDLVLSDPENYILKNKTSVLLICGNSDAGTCPEGYRCLKAGENDHGYTSFDSFAM 360
 DB 301 WESIDLVLSDPENYILKNKTSVLLICGNSDAGTCPEGYRCLKAGENDHGYTSFDSFAM 360
 QY 361 AFLALFRLMTQDCWRLYQOTLRSGAKTYMIFPMLVIFLGSFVYVILAVVANAAYEON 420
 DB 361 AFLALFRLMTQDCWRLYQOTLRSGAKTYMIFPMLVIFLGSFVYVILAVVANAAYEON 420

QY 421 QATIAETBEKEKRFQENAMEMLKKHEALITRGDVTVSRSLSLMSPLAPVNSHERSKRK 480
 Db 421 QATIAETBEKEKRFQENAMEMLKKHEALITRGDVTVSRSLSLMSPLAPVNSHERSKRK 480
 QY 481 RRSSTGTEEGEDRLPKSDSEDDPRAMNHLSTRGLSRTSMKPRSSRGSLFTTRRDDLGE 540
 Db 481 RRSSTGTEEGEDRLPKSDSEDDPRAMNHLSTRGLSRTSMKPRSSRGSLFTTRRDDLGE 540
 QY 541 ADFADENSTAGESHRTSLLPVMPLRRTSAQOGSPGTSAPGHALHKKNSJTYDCNVCV 600
 Db 541 ADFADENSTAGESHRTSLLPVMPLRRTSAQOGSPGTSAPGHALHKKNSJTYDCNVCV 600
 QY 601 VSLGAGDPEATSPGSHLLRPVMLBHPDPTTTPSEEGPGQMLTQAPCVDGFEERGAQ 660
 Db 601 VSLGAGDPEATSPGSHLLRPVMLBHPDPTTTPSEEGPGQMLTQAPCVDGFEERGAQ 660
 QY 661 PALSASVUTSALBEEBSRHKPCPCWNLQORYLIWECCLPMSIKOGVKLVMDPFTD 720
 Db 661 PALSASVUTSALBEEBSRHKPCPCWNLQORYLIWECCLPMSIKOGVKLVMDPFTD 720
 QY 721 LTTMCIYVNTLPMALHEHNTSEFEEMLOVGNLFTGIFTAEMTFKIIALDPYYPQOG 780
 Db 721 LTTMCIYVNTLPMALHEHNTSEFEEMLOVGNLFTGIFTAEMTFKIIALDPYYPQOG 780
 QY 781 WNIIPDSIIIVILSLMEIGLSRMSNLSVLRSFLLRPFKLAKSWPTNTLTIKIIGNSVGLG 840
 Db 781 WNIIPDSIIIVILSLMEIGLSRMSNLSVLRSFLLRPFKLAKSWPTNTLTIKIIGNSVGLG 840
 QY 841 NLTVLAIIVFIFAVVGQOLFQKNYSELKSDSGLLPRHMDPFHAFILIRIICGEMI 900
 Db 841 NLTVLAIIVFIFAVVGQOLFQKNYSELKSDSGLLPRHMDPFHAFILIRIICGEMI 900
 QY 901 ETMDMCEVSGSLCLLVFLVWVGNLVNLFALILSSFSANLTPADDERMNLQ 960
 Db 901 ETMDMCEVSGSLCLLVFLVWVGNLVNLFALILSSFSANLTPADDERMNLQ 960
 QY 961 LALARIORGLRFVKKTWDFCCGLRORPQKPAALAAQOLPSCIATPSPPPETEKVP 1020
 Db 961 LALARIORGLRFVKKTWDFCCGLRORPQKPAALAAQOLPSCIATPSPPPETEKVP 1020
 QY 1021 PTRKETRFEEBEGQPGQTPGDBEPVCPIAVASDTDDQEDDEENSLGTEBESSK-QESQ 1079
 Db 1021 PTRKETRFEEBEGQPGQTPGDBEPVCPIAVASDTDDQEDDEENSLGTEBESSK-QESQ 1079
 QY 1080 PVSQGPBARPPDSRTSQQVSATSSAABASASQADRWQKAPQAPGCGETPBDSCSGS 1139
 Db 1081 PVSQGPBARPPDSRTSQQVSATSSAABASASQADRWQKAPQAPGCGETPBDSCSGS 1140
 QY 1140 TADMNTAELLEQIPDLGQDVDPEDCFTBGCVRRCPCCAVDTTQAPGKVMRLKTCYH 1199
 Db 1141 TADMNTAELLEQIPDLGQDVDPEDCFTBGCVRRCPCCAVDTTQAPGKVMRLKTCYH 1200
 QY 1200 IVESHMPEFTFIIEMILSSGALAFEDIYLEBKTIKVLLEVADKMTYVVFVLEMLLKVA 1259
 Db 1201 IVESHMPEFTFIIEMILSSGALAFEDIYLEBKTIKVLLEVADKMTYVVFVLEMLLKVA 1260
 QY 1260 YGPKKYFTNANCMDFLIVDVSVLSVANTLGFAPMGPIKSLRTIARLRPRLALSREBGM 1319
 Db 1261 YGPKKYFTNANCMDFLIVDVSVLSVANTLGFAPMGPIKSLRTIARLRPRLALSREBGM 1320
 QY 1320 RVVNAVALVGAIPSIINAVLVCILFPLISIMGVNLFAKFGRCINOTEGDPLANTTYNN 1379
 Db 1321 RVVNAVALVGAIPSIINAVLVCILFPLISIMGVNLFAKFGRCINOTEGDPLANTTYNN 1380
 QY 1380 KSQCESLNLGSELVYTKVKNPNDVAGYALALQVATFKGWNMDIYAAVDSRGYEEQOW 1439
 Db 1381 KSQCESLNLGSELVYTKVKNPNDVAGYALALQVATFKGWNMDIYAAVDSRGYEEQOW 1440
 QY 1440 EYNLYMYTYFYFIIRGSEFTLNLFTGVTINFNQOKKGGODIFMTEBQKTYNANKK 1499
 Db 1441 EYNLYMYTYFYFIIRGSEFTLNLFTGVTINFNQOKKGGODIFMTEBQKTYNANKK 1500
 QY 1500 LGSKKPQKPIPRPLNKYGFIIDYVTKOAFDVTIMFLICLNMVTMMVETDQSPKINIL 1559

Db 1501 LGSKKPQKPIPRPLNKYGFIIDYVTKOAFDVTIMFLICLNMVTMMVETDQSPKINIL 1560
 QY 1560 AKINILFAIFTEGECIVLAAALRHYYFTNSWNIPOPVVILISIVGVUSDIIOKYFESFT 1619
 Db 1561 AKINILFAIFTEGECIVLAAALRHYYFTNSWNIPOPVVILISIVGVUSDIIOKYFESFT 1620
 QY 1620 LFRVIRLARIGRIILIRIGAKGIRTLFALMMSLALFNIGLLFVMEFYISIFGMANPA 1679
 Db 1621 LFRVIRLARIGRIILIRIGAKGIRTLFALMMSLALFNIGLLFVMEFYISIFGMANPA 1680
 QY 1680 YKMEAGIDDMENFOTFANSMCLFQITTSAGMDGLSPIINTGPPYCDPTLPNSNGSRG 1739
 Db 1681 YKMEAGIDDMENFOTFANSMCLFQITTSAGMDGLSPIINTGPPYCDPTLPNSNGSRG 1740
 QY 1740 DQSPAVGILPFTTYIIISFLIVNMVYAIILENSVAETESTELSEDDPFMEFYIWEK 1799
 Db 1741 DQSPAVGILPFTTYIIISFLIVNMVYAIILENSVAETESTELSEDDPFMEFYIWEK 1800
 QY 1800 FDPKATQFTEYSVLSDFADALSEPLRIAKPNOISLINDLPMVSGDRICHMDILFAFTKR 1859
 Db 1801 FDPKATQFTEYSVLSDFADALSEPLRIAKPNOISLINDLPMVSGDRICHMDILFAFTKR 1860
 QY 1860 VLGSSEMDALKIOMEKFMANPSKISYBPIITTLRKHEVSAWVQRAFRRHLQRS 1919
 Db 1861 VLGSSEMDALKIOMEKFMANPSKISYBPIITTLRKHEVSAWVQRAFRRHLQRS 1920
 QY 1920 LKHSFLEFROQAGSLSEEDA PERBGLIAYVMSNFSPCLGPPSSSISSTSPSSYSV 1979
 Db 1921 LKHSFLEFROQAGSLSEEDA PERBGLIAYVMSNFSPCLGPPSSSISSTSPSSYSV 1980
 QY 1980 TRATSNDLQVNGSDVSHSEDLADPPSPDRRESTIV 2015
 Db 1981 TRATSNDLQVNGSDVSHSEDLADPPSPDRRESTIV 2016
 RESULT 14
 AAB82245
 ID AAB82245 standard; protein: 2016 AA.
 XX AAB82245;
 AC 21-JUN-2001 (first entry)
 DT 21-JUN-2001 (first entry)
 DE Human SCN5A mutant S1787N.
 DE SCN5A; long QT syndrome; LQTS; cardiovascular disease;
 KW Romano-Ward syndrome; diagnosis; prognosis; therapy; drug screening;
 KW mutant; mutein.
 XX Homo sapiens.
 OS Homo sapiens.
 PN WO200124681-A2.
 PD 12-APR-2001.
 PF 09-AUG-2000; 2000WO-US021660.
 PR 09-AUG-1999; 99US-0147486P.
 PR 17-MAR-2000; 2000US-0190057P.
 PA (UTAH) UNIV UTAH RES FOUND.
 XX Keating MT, Splawski I;
 PI WPI; 2001-290564/30.
 XX New XVLQRT and SCN5A genes, which contains alterations or mutations,
 PT useful in diagnostic/prognostic or drug screening methods, particularly in
 PT mutation analyses for screening individuals with or at risk for long QT
 syndrome.
 PS Claim 31; Page; 76pp; English.

XX The present sequence is that of the claimed S1787N mutant of the human
CC SCNSA protein. The mutant is encoded by an SCNSA mutant gene in which a
CC G/A mutation alters codon 1787 from AGT to AAT. Mutations of the SCNSA
CC gene are implicated in Romano-Ward syndrome, the autosomal dominant form
CC of Long QT syndrome (LQTS). Mutations newly discovered in the SCNSA gene
CC lead to the following amino acid alterations in the encoded protein:
CC D114N, I1501V, delP1617, R1623L, E1784K and S1787N. Knowledge of the
CC mutations provides means for assessing a risk in a human subject for
CC LQTS, for diagnosing a mutation which causes LQTS, and for screening for
CC drugs useful in treating a human having a mutation in the SCNSA gene.
CC Note: The present sequence is not shown in the specification but is
CC derived from the KVTQT-1 sequence given in the Sequence Listing (see
CC AAB82220)
CC
CC
SQ Sequence 2016 AA:

Query Match 99.4%; Score 10429.5; DB 4; Length 2016;
Best Local Similarity 99.5%; Pred. No. 0;
Matches 2005; Conservative 3; Mismatches 7; Indels 1; Gaps 1;

QY 1 MANFLPRGTSFRFRFTRESLAIEKMAEKQARGSTTLQESREGLPREEAPRQDLQA 60
Db 1 MANFLPRGTSFRFRFTRESLAIEKMAEKQARGSTTLQESREGLPREEAPRQDLQA 60
QY 61 SKKLPLDYGNDPQELIGEPLEDDLPFYSTQKTFIVLNGKTI FRPSATNALVYSPFPI 120
Db 61 SKKLPLDYGNDPQELIGEPLEDDLPFYSTQKTFIVLNGKTI FRPSATNALVYSPFPI 120
QY 121 RRAVKILVHSLFNNLIMCTILTCNVMAOHDPPEWTXYVEYTFPAITYPSLVYKILARG 180
Db 121 RRAVKILVHSLFNNLIMCTILTCNVMAOHDPPEWTXYVEYTFPAITYPSLVYKILARA 180
QY 181 FCLHAFTELRDPWNNLIDPSVILIMAYTTEFVDLGNVSALRTRFRVLRALKTISVIGLKTIV 240
Db 181 FCLHAFTELRDPWNNLIDPSVILIMAYTTEFVDLGNVSALRTRFRVLRALKTISVIGLKTIV 240
QY 241 GALIOSVKKLADVMVLTFCISVFPALIGLFMGULRHKCVNFPALMNGTNGSVADGLV 300
Db 241 GALIOSVKKLADVMVLTFCISVFPALIGLFMGULRHKCVNFPALMNGTNGSVADGLV 300
QY 301 MESLDLYSDPENYLLKNGTSDVLLCGNSSDAGTPEGYRCIKAGENPDHGYTSPDFAM 360
Db 301 MESLDLYSDPENYLLKNGTSDVLLCGNSSDAGTPEGYRCIKAGENPDHGYTSPDFAM 360
QY 361 AFLALFRIMTODCWERLYQOTLRSAKTYMIFPMLVIFLGSFYLVNLLAVVAMAYEON 420
Db 361 AFLALFRIMTODCWERLYQOTLRSAKTYMIFPMLVIFLGSFYLVNLLAVVAMAYEON 420
QY 421 QATTIETBEKEKRFQAMEMLKKEHEALTRGVDTVVSRLSLEMSFLAPNHSHERSKRK 480
Db 421 QATTIETBEKEKRFQAMEMLKKEHEALTRGVDTVVSRLSLEMSFLAPNHSHERSKRK 480
QY 481 RMSSTEEGEGDRLPKSDSDGPRAMNHLSTFRGLSRTSMKPRSRGSIFFRRRDLSE 540
Db 481 RMSSTEEGEGDRLPKSDSDGPRAMNHLSTFRGLSRTSMKPRSRGSIFFRRRDLSE 540
QY 541 ADFADENSTAGESHRTSLVWPRLRRTSAQGPSPTGSAFGHALGKXSTYDCNGV 600
Db 541 ADFADENSTAGESHRTSLVWPRLRRTSAQGPSPTGSAFGHALGKXSTYDCNGV 600
QY 601 VSLIAGDPPEATSPGSHLIRPYMLBHPDTPPTPSEBPGAPQWLTQAPCVDFEPEGARQ 660
Db 601 VSLIAGDPPEATSPGSHLIRPYMLBHPDTPPTPSEBPGAPQWLTQAPCVDFEPEGARQ 660
QY 661 RLASAVSVLTSLAELEBSRHKCPQCMNLAQRYLIWECCLMMSIKQGVKLVNMDPFD 720
Db 661 RLASAVSVLTSLAELEBSRHKCPQCMNLAQRYLIWECCLMMSIKQGVKLVNMDPFD 720
QY 721 LTIITACIVANTLFMALEHNMTSEFEMLQVGNLVTGTIFTAEMTFKIIALDPYYFPQG 780
Db 721 LTIITACIVANTLFMALEHNMTSEFEMLQVGNLVTGTIFTAEMTFKIIALDPYYFPQG 780

QY 781 WNTFDSITVTLISMEGLSRMSNLVYLRFLRLRVFLAKSMPNTLTKIIGNSVGALG 840
Db 781 WNTFDSITVTLISMEGLSRMSNLVYLRFLRLRVFLAKSMPNTLTKIIGNSVGALG 840
QY 841 NLTIVLAIIVFPAVGMQLFGKXYSRLSDSGLLPRWMDPFHAFLIIFRILGEMI 900
Db 841 NLTIVLAIIVFPAVGMQLFGKXYSRLSDSGLLPRWMDPFHAFLIIFRILGEMI 900
QY 901 ETWMDCHVEVGQSICLVFLVNWIGLVYVNLFLMILLSSFSADNLTADDERNNLQ 960
Db 901 ETWMDCHVEVGQSICLVFLVNWIGLVYVNLFLMILLSSFSADNLTADDERNNLQ 960
QY 961 LALARIQRLRFVKRTWDCCGLLRQRPKAPALAAQQLPSCIATPYSPPEETKVP 1020
Db 961 LALARIQRLRFVKRTWDCCGLLRQRPKAPALAAQQLPSCIATPYSPPEETKVP 1020
QY 1021 PTRKETRFEEGEOQGTDPDPEVCYPIVAESDTDDOEDEBNSIGTEBESSK-OESQ 1079
Db 1021 PTRKETRFEEGEOQGTDPDPEVCYPIVAESDTDDOEDEBNSIGTEBESSK-OESQ 1080
QY 1080 PVSGGPEAPDPDSRTWSQVSAATASEAASASQADWRQWKAEPQAPCCGCTPEDESCSGS 1139
Db 1081 PVSGGPEAPDPDSRTWSQVSAATASEAASASQADWRQWKAEPQAPCCGCTPEDESCSGS 1140
QY 1140 TADMTNTAAILLEQIPDLGQDVKDPEDCFEGCVARCCCAVDTTQAPGKYWMRLRKTQYH 1199
Db 1141 TADMTNTAAILLEQIPDLGQDVKDPEDCFEGCVARCCCAVDTTQAPGKYWMRLRKTQYH 1200
QY 1200 IVEHSWETFLIFMILLSSGALAFEDYLEBKTIKVLLEYADMFTYVFLVLEMLKVA 1259
Db 1201 IVEHSWETFLIFMILLSSGALAFEDYLEBKTIKVLLEYADMFTYVFLVLEMLKVA 1260
QY 1260 YGFKKYFTTNAMCMLDELIVDSVLSVYANTLGFPEMCPISIRTLRLRPRALSRREGM 1319
Db 1261 YGFKKYFTTNAMCMLDELIVDSVLSVYANTLGFPEMCPISIRTLRLRPRALSRREGM 1320
QY 1320 RYVYNALVGAIPSIIMNVLVCLIFMLIFSLMGVLFAGKTRCINOTEGDLPLNYTIVNN 1379
Db 1321 RYVYNALVGAIPSIIMNVLVCLIFMLIFSLMGVLFAGKTRCINOTEGDLPLNYTIVNN 1380
QY 1380 KSQCESLNTGELYWTKVNFVNDVAGVYALLOVATFKGMDIMYAAVDSRGYEBOPQW 1439
Db 1381 KSQCESLNTGELYWTKVNFVNDVAGVYALLOVATFKGMDIMYAAVDSRGYEBOPQW 1440
QY 1440 EYNYTMYTPIYTFIRBSFPTLNFIFGVYIIDNENQKKKGGODIEMTEBOKKYNNAMKK 1499
Db 1441 EYNYTMYTPIYTFIRBSFPTLNFIFGVYIIDNENQKKKGGODIEMTEBOKKYNNAMKK 1500
QY 1500 LGSKKPOKPIPRPLNKYQGFIFDIYTRQAPDVTIMPLICLNMVTMMVETDDQSEKINIIL 1559
Db 1501 LGSKKPOKPIPRPLNKYQGFIFDIYTRQAPDVTIMPLICLNMVTMMVETDDQSEKINIIL 1560
QY 1560 AKINLLFVALFTGECIVKLAALRHYFTNSWNIFDPAVVYIISIVGYLSDITIQKTFSPPT 1619
Db 1561 AKINLLFVALFTGECIVKLAALRHYFTNSWNIFDPAVVYIISIVGYLSDITIQKTFSPPT 1620
QY 1620 LFRVYIRLARIGRIIRLRGAGGITTLFALMMSIPALFNIGLLFLVWFYISIGMANFA 1679
Db 1621 LFRVYIRLARIGRIIRLRGAGGITTLFALMMSIPALFNIGLLFLVWFYISIGMANFA 1680
QY 1680 YVKNBAGIDDMENFOTFANSMLCLFOITTSAGMDGLSPILANTGPYCDPTLPNSNGSRG 1739
Db 1681 YVKNBAGIDDMENFOTFANSMLCLFOITTSAGMDGLSPILANTGPYCDPTLPNSNGSRG 1740
QY 1740 DCGSPAVGILFFTYIIISFLIVNMVYIAILLENFSVATEESTPLENDEDDMEYEIWEK 1799
Db 1741 DCGSPAVGILFFTYIIISFLIVNMVYIAILLENFSVATEESTPLENDEDDMEYEIWEK 1800
QY 1800 FDPPEATQTIERYSVLSDFPDALSEPLRLAKPQOISLIMNDLPMVSGDRTHCDDILPAFTR 1859
Db 1801 FDPPEATQTIERYSVLSDFPDALSEPLRLAKPQOISLIMNDLPMVSGDRTHCDDILPAFTR 1860
QY 1860 VLGSBGENDALKIQWEEKFMANPSKISYEDITTLTRKHEEVSAMVIGQAFRRHLQORS 1919

|||||
DB 1861 VLGESEMDALKIOMEKEMANPSKISYEPIITTLRRGHEVSAMVIOARFRRLQGS 1920
QY 1920 LKHAFLFRQOQSGSLSEEDAPEREGLIAYVMSKPSRPLGPPSSSISSTSPSYOSV 1979
DB 1921 LKHAFLFRQOQSGSLSEEDAPEREGLIAYVMSKPSRPLGPPSSSISSTSPSYOSV 1980
QY 1980 TRATSDNLQVRGSDYSHSEDLADPPSPDRDRESIV 2015
DB 1981 TRATSDNLQVRGSDYSHSEDLADPPSPDRDRESIV 2016

RESULT 15
AAB82241
ID AAB82241 standard; protein; 2016 AA.
AC AAB82241;
XX 21-JUN-2001 (first entry)
DT 21-JUN-2001 (first entry)
XX Human SCNSA mutant L1501V.
DE SCNSA; Long QT syndrome; LQTS; cardiovascular disease;
KM Romano-Ward syndrome; diagnosis; prognosis; therapy; drug screening;
KW mutant; mte1n.
XX Homo sapiens.
OS WO200124681-A2.
PN 12-APR-2001.
XX 09-AUG-2000; 2000MO-US021660.
PD 09-AUG-1999; 99US-0147488P.
XX 17-MAR-2000; 2000US-0190057P.
PR (UTAH) UNIV UTAH RES FOUND.
XX Keating MT, Splawski I;
PI WPI; 2001-290564/30.
DR New KVLQ1 and SCNSA genes, which contains alterations or mutations.
XX useful in diagnostic/prognostic or drug screening methods, particularly in
PT mutational analyses for screening individuals with or at risk for long QT
PT syndrome.
XX Claim 31; Page; 76pp; English.
PS
XX The present sequence is that of the claimed L1501V mutant of the human
CC SCNSA protein. The mutant is encoded by an SCNSA mutant gene in which a
CC C/G mutation alters codon 1501 from CTG to GTG. Mutations of the SCNSA
CC gene are implicated in Romano-Ward syndrome, the autosomal dominant form
CC of long QT syndrome (LQTS). Mutations newly discovered in the SCNSA gene
CC lead to the following amino acid alterations in the encoded protein:
CC D114N, L1501V, del1617, R1623L, E1784K and S1787N. Knowledge of the
CC mutations provides means for assessing a risk in a human subject for
CC LQTS, for diagnosing a mutation which causes LQTS, and for screening for
CC drugs useful in treating a human having a mutation in the SCNSA gene.
CC Note: The present sequence is not shown in the specification but is
CC derived from the KVLQ1-1 sequence given in the Sequence Listing (see
CC AAB82220)
XX
SQ Sequence 2016 AA;

Query Match 99.4%; Score 10429.5; DB 4; Length 2016;
Best Local Similarity 99.5%; Pred. No. 0;
Matches 2005; Conservative 3; Mismatches 7; Indels 1; Gaps 1;

QY 61 SKGLPDLGNPQOELIGBPLEDLDPFYSTOKTFIVLNGKTIIFRSATNALVYLSFHHI 120
DB 61 SKGLPDLGNPQOELIGBPLEDLDPFYSTOKTFIVLNGKTIIFRSATNALVYLSFHHI 120
QY 121 RRAAVKILVHSLFNMLINCTIITNCVMAQHDPPWTKYVEYTFALYTESLVKILANG 180
DB 121 RRAAVKILVHSLFNMLINCTIITNCVMAQHDPPWTKYVEYTFALYTESLVKILANG 180
QY 181 FCLHAFYLRDPWNLADPSVILIMAYTEFVLDGANVSALRTFVLAALKTISVIGLKTIV 240
DB 181 FCLHAFYLRDPWNLADPSVILIMAYTEFVLDGANVSALRTFVLAALKTISVIGLKTIV 240
QY 241 GALLISVKKLADVWVITVFCLSVFALIGLOLFMGNLRRKCVNFTALNGTNGSVADGLIV 300
DB 241 GALLISVKKLADVWVITVFCLSVFALIGLOLFMGNLRRKCVNFTALNGTNGSVADGLIV 300
QY 301 WESLDLYLSDPENNYLLKNGTSDVLLCGNSSDAGTCPEGYRCLKAGENDHGYTSPDSFAM 360
DB 301 WESLDLYLSDPENNYLLKNGTSDVLLCGNSSDAGTCPEGYRCLKAGENDHGYTSPDSFAM 360
QY 361 AFLALFRIMTQDCWERYOQTLRSAGKIYMTFPMVLVILGSRFYLVNLLAVAMAYEEN 420
DB 361 AFLALFRIMTQDCWERYOQTLRSAGKIYMTFPMVLVILGSRFYLVNLLAVAMAYEEN 420
QY 421 QATTAEFEKEXRPOEAMEMLKKEHEALTTRGVDTVSRSSLEMSPLAPVNSHERSKRX 480
DB 421 QATTAEFEKEXRPOEAMEMLKKEHEALTTRGVDTVSRSSLEMSPLAPVNSHERSKRX 480
QY 481 RMSSGTECEGDRLPKSDSEDPGRAMNHLSTLRGLSRTSMKRRSSRGSIFFTRRRDLGSE 540
DB 481 RMSSGTECEGDRLPKSDSEDPGRAMNHLSTLRGLSRTSMKRRSSRGSIFFTRRRDLGSE 540
QY 541 ADPADDENSTAGESHSRTSLVWPPLRTSAQOGPSGTSAFGALHGKKNSTYDCNIV 600
DB 541 ADPADDENSTAGESHSRTSLVWPPLRTSAQOGPSGTSAFGALHGKKNSTYDCNIV 600
QY 601 VSLGAGPBPATSPESHLLRPVMLEHPDPTTSPSEBPQOMLTSQACVDFEFGARQ 660
DB 601 VSLGAGPBPATSPESHLLRPVMLEHPDPTTSPSEBPQOMLTSQACVDFEFGARQ 660
QY 661 PALSASVYLTALBELLESRRHKCPQNRLAQRYLIWECPLMWSIKOGVKLVVMDPFTD 720
DB 661 PALSASVYLTALBELLESRRHKCPQNRLAQRYLIWECPLMWSIKOGVKLVVMDPFTD 720
QY 721 LFTTICIVLNTLPMALHEYNTSEPEMLQVGNLVFTGIPTAEMTFKIIALDPYYFOQG 780
DB 721 LFTTICIVLNTLPMALHEYNTSEPEMLQVGNLVFTGIPTAEMTFKIIALDPYYFOQG 780
QY 781 WNIIFDSIIIVILSILMEIGSRMSNLVLSRPFLLRVFKLAKSMPTLNTLIIKIIGNSVGALG 840
DB 781 WNIIFDSIIIVILSILMEIGSRMSNLVLSRPFLLRVFKLAKSMPTLNTLIIKIIGNSVGALG 840
QY 841⁴NLTIVLAIIVIPAVVGQQLPGKNYSRLRDSGLIPRHHMDFFHAFLIIFRILGEMI 900
DB 841 NLTIVLAIIVIPAVVGQQLPGKNYSRLRDSGLIPRHHMDFFHAFLIIFRILGEMI 900
QY 901 ETMDQCEVSGQSLCLLVLLVWVIGNLVNLFLIALILSSPSADNLTAPDEBRMNIQ 960
DB 901 ETMDQCEVSGQSLCLLVLLVWVIGNLVNLFLIALILSSPSADNLTAPDEBRMNIQ 960
QY 961 LALARIQGLRFVVRTTMDPCCGILLRORPOKPAALAAAGOLPSCATATYSPPEPTERYP 1020
DB 961 LALARIQGLRFVVRTTMDPCCGILLRORPOKPAALAAAGOLPSCATATYSPPEPTERYP 1020
QY 1021 PTRKETPEEBEQGCGIIPDPEVCVPIAAESDTDDQDEDENSLGTBESSKQGSQ 1080
DB 1021 PTRKETPEEBEQGCGIIPDPEVCVPIAAESDTDDQDEDENSLGTBESSKQGSQ 1080
QY 1081 PVSQGPAPPDPSRTWSQVSATASSEAEASASQADWRQWKAEPQAPGCGETPEBSCSGS 1139
DB 1081 PVSQGPAPPDPSRTWSQVSATASSEAEASASQADWRQWKAEPQAPGCGETPEBSCSGS 1140

QY 1140 TADMTNTAELLQIIDLGDVYKDEDECTEGCVRRCPCCAVDTTQAPGKTMRLKTCYH 1199
 DB 1141 TADMTNTAELLQIIDLGDVYKDEDECTEGCVRRCPCCAVDTTQAPGKTMRLKTCYH 1200
 QY 1200 IVEHSWEPFTIIFMILLSSGALAPEDYLERKTIKYLLREXADKMFYVFLKMLKVA 1259
 DB 1201 IVEHSWEPFTIIFMILLSSGALAPEDYLERKTIKYLLREXADKMFYVFLKMLKVA 1260
 QY 1260 YGFKKYFTNACWMLDFLVDVSLVSVANTLGAEMGPISKIRTLRALRPLRALSREGM 1319
 DB 1261 YGFKKYFTNACWMLDFLVDVSLVSVANTLGAEMGPISKIRTLRALRPLRALSREGM 1320
 QY 1320 RYVNAVALVGAIPSIINVLVLCILFWLIFSIINGVNLPAKFERCINQTEBGLPLATTTVNN 1379
 DB 1321 RYVNAVALVGAIPSIINVLVLCILFWLIFSIINGVNLPAKFERCINQTEBGLPLATTTVNN 1380
 QY 1380 KSQCESLNLTEGLVYTKKYNPDNAGAGYLLALQVATKGMMDIYAAVDSRGYBOPQW 1439
 DB 1381 KSQCESLNLTEGLVYTKKYNPDNAGAGYLLALQVATKGMMDIYAAVDSRGYBOPQW 1440
 QY 1440 EYNLYMYIYFVFIIFGSEFTLNLPIGYIIDNFQOKKLGQODIEMTEBQKXYNAMKX 1499
 DB 1441 EYNLYMYIYFVFIIFGSEFTLNLPIGYIIDNFQOKKLGQODIEMTEBQKXYNAMKX 1500
 QY 1500 LGSKKPKQPIPRPLNKYOGFIIDYTKQAFDVTIMFLICLANNVTMMETDQSPKINIL 1559
 DB 1501 LGSKKPKQPIPRPLNKYOGFIIDYTKQAFDVTIMFLICLANNVTMMETDQSPKINIL 1560
 QY 1560 AKINILFVAIFEGECIVLAALRHYEFNSNNIPFVVVYLSIVGTVSDIIOKFFSPPT 1619
 DB 1561 AKINILFVAIFEGECIVLAALRHYEFNSNNIPFVVVYLSIVGTVSDIIOKFFSPPT 1620
 QY 1620 LFRVIRLARIGRIILIRIGAKGIRTLFPALMWSLPALFNIGLLPLVNFYISIFGMANPA 1679
 DB 1621 LFRVIRLARIGRIILIRIGAKGIRTLFPALMWSLPALFNIGLLPLVNFYISIFGMANPA 1680
 QY 1680 YVKEWAGIDDMENFOTFANSMLCLFOITTSAGWDGLSPILNTGPPYCDPTLPNSNGSRG 1739
 DB 1681 YVKEWAGIDDMENFOTFANSMLCLFOITTSAGWDGLSPILNTGPPYCDPTLPNSNGSRG 1740
 QY 1740 DCGSPAVGLFETTTTIIISFLVNNMYTALILENFSVATEESTEPLEDDPFMEIEMK 1799
 DB 1741 DCGSPAVGLFETTTTIIISFLVNNMYTALILENFSVATEESTEPLEDDPFMEIEMK 1800
 QY 1800 PDPEATQFIIEYVLSDFADALSEPLRIAKPNQISILINDLPVWSGDRJHCMDILFAFTKR 1859
 DB 1801 PDPEATQFIIEYVLSDFADALSEPLRIAKPNQISILINDLPVWSGDRJHCMDILFAFTKR 1860
 QY 1860 VLGESEMDALKIOMEKFMANPSKISTEPIITTLRRKHEVSAMVIOARFRHLQRS 1919
 DB 1861 VLGESEMDALKIOMEKFMANPSKISTEPIITTLRRKHEVSAMVIOARFRHLQRS 1920
 QY 1920 LKHAASLFRQOAGSGISEEDAPERBGLIYVNSSENRPLGPPSSSISSTSPSYDSV 1979
 DB 1921 LKHAASLFRQOAGSGISEEDAPERBGLIYVNSSENRPLGPPSSSISSTSPSYDSV 1980
 QY 1980 TRATSNDLQVRGSDYSHSEDLADFPSPPRDRESIV 2015
 DB 1981 TRATSNDLQVRGSDYSHSEDLADFPSPPRDRESIV 2016

KW mutant; mutein.
 XX Homo sapiens.
 OS WO200124681-A2.
 PN 12-APR-2001.
 PD 09-AUG-2000; 2000MO-US021660.
 PF 09-AUG-1999; 99US-0147488P.
 PR 17-MAR-2000; 2000US-0190057P.
 XX (UTAH) UNIV UTAH RES FOUND.
 PA Keating MT, Splawski I;
 PI WPI; 2001-290564/30.
 DR New KYLQRT and SCN5A gene, which contains alterations or mutations,
 XX useful in diagnostic/prognostic or drug screening methods, particularly in
 PT mutational analyses for screening individuals with or at risk for long QT
 PT syndrome.
 PS Claim 31; Page; 76pp; English.
 CC The present sequence is that of the claimed E1784K mutant of the human
 CC SCN5A protein. The mutant is encoded by an SCN5A mutant gene in which a
 CC G/A mutation alters codon 1784 from GAG to AAG. Mutations of the SCN5A
 CC gene are implicated in Romano-Ward syndrome, the autosomal dominant form
 CC of long QT syndrome (LQTS). Mutations newly discovered in the SCN5A gene
 CC lead to the following amino acid alterations in the encoded protein:
 CC D1114N, L1501V, delF1617, R1623L, E1784K and S1787N. Knowledge of the
 CC mutations provides means for assessing a risk in a human subject for
 CC LQTS, for diagnosing a mutation which causes LQTS, and for screening for
 CC drugs useful in treating a human having a mutation in the SCN5A gene.
 CC Note: The present sequence is not shown in the specification but is
 CC derived from the KYLQRT-1 sequence given in the Sequence Listing (see
 CC AAB82220)
 CC
 SQ Sequence 2016 AA;
 Query Match 99.4%; Score 10428.5; DB 4; Length 2016;
 Best local similarity 99.5%; Pred. No. 0;
 Matches 2005; Conservative 3; Mismatches 7; Indels 1; Gaps 1;
 QY 1 MANFLPRGTSSFFRRFTRESIALEKMAEKQARGSTTLOESREGLEPEEAPRQDLQA 60
 DB 1 MANFLPRGTSSFFRRFTRESIALEKMAEKQARGSTTLOESREGLEPEEAPRQDLQA 60
 QY 61 SKKLPLDLYGNPDELIGEPLEDLPFYSTQKTFIVLNKKGKTIFFFSATNLVYLSPPHPI 120
 DB 61 SKKLPLDLYGNPDELIGEPLEDLPFYSTQKTFIVLNKKGKTIFFFSATNLVYLSPPHPI 120
 QY 121 RRAAVKILVHSLFNNLIMCTILTNCFVMAQDPPPMTKYVYFTTAYTFESLVKILAR 180
 DB 121 RRAAVKILVHSLFNNLIMCTILTNCFVMAQDPPPMTKYVYFTTAYTFESLVKILAR 180
 QY 181 FCLHAFTEFLRDPNMWMLDSESVIIMAYTTEFVDLGNVSALRFRVRLPAKTIISVIGLKTIV 240
 DB 181 FCLHAFTEFLRDPNMWMLDSESVIIMAYTTEFVDLGNVSALRFRVRLPAKTIISVIGLKTIV 240
 QY 241 GALIOSVKKLADVWVLYTCFISVAPALLGLQIFMKNLIRHKCVRNFTALNGTNGSVYADGLV 300
 DB 241 GALIOSVKKLADVWVLYTCFISVAPALLGLQIFMKNLIRHKCVRNFTALNGTNGSVYADGLV 300
 QY 301 WESLDLYSDPENYLLKNGTSDVLLCGNSSDAGTCPEEGYRCLKAGENDHGYTSFDSFAM 360
 DB 301 WESLDLYSDPENYLLKNGTSDVLLCGNSSDAGTCPEEGYRCLKAGENDHGYTSFDSFAM 360
 QY 361 AFLALFRIMTODCWERLYQOTLRSAKITYMFMLVIFLGSFYVNLILAVANAYEERON 420
 DB 361 AFLALFRIMTODCWERLYQOTLRSAKITYMFMLVIFLGSFYVNLILAVANAYEERON 420

QY	421	QATI	KETER	KER	POE	AMEM	KKE	HEAL	TI	RG	VD	VSS	SU	MS	GLA	PVN	SHER	KRR	480	
Db	421	QATI	KETER	KER	POE	AMEM	KKE	HEAL	TI	RG	VD	VSS	SU	MS	GLA	PVN	SHER	KRR	480	
QY	481	RMS	STEE	CE	GE	R	L	P	K	S	D	E	D	E	P	R	A	M	NH	540
Db	481	RMS	STEE	CE	GE	R	L	P	K	S	D	E	D	E	P	R	A	M	NH	540
QY	541	AD	PAD	EN	STAGE	SE	SH	RTS	LL	V	P	M	L	R	T	S	A	O	G	600
Db	541	AD	PAD	EN	STAGE	SE	SH	RTS	LL	V	P	M	L	R	T	S	A	O	G	600
QY	601	VS	LL	GAG	PE	AT	S	P	SH	L	R	P	M	L	E	H	N	T	S	660
Db	601	VS	LL	GAG	PE	AT	S	P	SH	L	R	P	M	L	E	H	N	T	S	660
QY	661	RA	LS	AV	S	VL	T	S	L	B	E	S	R	H	K	C	P	C	M	720
Db	661	RA	LS	AV	S	VL	T	S	L	B	E	S	R	H	K	C	P	C	M	720
QY	721	LT	IT	MC	I	VL	NT	L	F	M	L	E	H	N	T	S	E	E	M	780
Db	721	LT	IT	MC	I	VL	NT	L	F	M	L	E	H	N	T	S	E	E	M	780
QY	781	W	N	I	P	S	I	I	V	I	S	I	M	E	L	G	S	R	M	840
Db	781	W	N	I	P	S	I	I	V	I	S	I	M	E	L	G	S	R	M	840
QY	841	N	L	T	I	V	L	A	I	I	V	F	P	A	V	A	W	G	M	900
Db	841	N	L	T	I	V	L	A	I	I	V	F	P	A	V	A	W	G	M	900
QY	901	E	T	M	D	C	M	E	V	S	G	S	L	C	L	I	F	L	V	960
Db	901	E	T	M	D	C	M	E	V	S	G	S	L	C	L	I	F	L	V	960
QY	961	L	A	L	A	R	I	O	R	G	L	R	P	V	K	T	T	M	D	1020
Db	961	L	A	L	A	R	I	O	R	G	L	R	P	V	K	T	T	M	D	1020
QY	1021	P	T	R	K	E	T	R	E	E	B	H	O	P	O	G	T	P	D	1080
Db	1021	P	T	R	K	E	T	R	E	E	B	H	O	P	O	G	T	P	D	1080
QY	1080	P	V	S	G	G	E	A	P	P	S	R	T	M	S	O	V	S	A	1140
Db	1081	P	V	S	G	G	E	A	P	P	S	R	T	M	S	O	V	S	A	1140
QY	1140	T	A	D	M	N	T	A	L	E	L	E	O	I	P	D	L	G	O	1199
Db	1141	T	A	D	M	N	T	A	L	E	L	E	O	I	P	D	L	G	O	1199
QY	1200	I	V	E	H	S	W	E	F	T	I	I	F	M	L	S	S	G	A	1259
Db	1201	I	V	E	H	S	W	E	F	T	I	I	F	M	L	S	S	G	A	1259
QY	1260	Y	G	F	K	K	Y	P	N	A	C	M	W	I	D	F	L	V	D	1319
Db	1261	Y	G	F	K	K	Y	P	N	A	C	M	W	I	D	F	L	V	D	1319
QY	1320	R	V	V	N	A	L	V	G	A	I	P	S	I	M	N	V	L	A	1379
Db	1321	R	V	V	N	A	L	V	G	A	I	P	S	I	M	N	V	L	A	1379
QY	1380	K	S	O	C	E	S	L	N	I	T	G	E	L	Y	T	K	V	N	1439
Db	1381	K	S	O	C	E	S	L	N	I	T	G	E	L	Y	T	K	V	N	14

QY	1500	IGSKKKQKQPIIRPLANKQGFEDIVTQQAPOVUTIMPLICLANNVTMMVETDQOSEKINIL	1559
Db	1501	LGSKKKQKQPIIRPLANKQGFEDIVTQQAPOVUTIMPLICLANNVTMMVETDQOSEKINIL	1560
QY	1560	AKINLLFAAITGBCIVLAAALRRHYXTNSWNIEDFVVVLISYGTVLSDIILQKYFSPPT	1619
Db	1561	AKINLLFAAITGBCIVLAAALRRHYXTNSWNIEDFVVVLISYGTVLSDIILQKYFSPPT	1620
QY	1620	LFRVIRLARIGRIILRLIRGAKGIRFTLLPALMMSIPALFNIGLLFLVMFYISIFGMANFA	1679
Db	1621	LFRVIRLARIGRIILRLIRGAKGIRFTLLPALMMSIPALFNIGLLFLVMFYISIFGMANFA	1680
QY	1680	YVKWEAGIDDMFNQOTRANSMCLCFQTTTSAGMGGLSPLINTGPPICDPTLPMSNSRG	1739
Db	1681	YVKWEAGIDDMFNQOTRANSMCLCFQTTTSAGMGGLSPLINTGPPICDPTLPMSNSRG	1740
QY	1740	DCGSPAVGILFFETTYIIISFLIYVNMVIAIILENFVSATBESTEPLSEDDPDMFYELMEK	1799
Db	1741	DCGSPAVGILFFETTYIIISFLIYVNMVIAIILENFVSATBESTEPLSEDDPDMFYELMEK	1800
QY	1800	FDPEATQPIEYSVLSDPADALSEPLRIAKPNQISLIMMDLPMVSGDRIHCMIDILFAFTR	1859
Db	1801	FDPEATQPIEYSVLSDPADALSEPLRIAKPNQISLIMMDLPMVSGDRIHCMIDILFAFTR	1860
QY	1860	VLGSGSEMDALKIOMEKFMANPSKISYEPIITTLRKKEEVSAMVIOQAFRRHLQORS	1919
Db	1861	VLGSGSEMDALKIOMEKFMANPSKISYEPIITTLRKKEEVSAMVIOQAFRRHLQORS	1920
QY	1920	LKHASPLFROQAGSGLSEBDAPEREGILAYVMSSENFSPRGPPSSSSISSTSPPSYDSV	1979
Db	1921	LKHASPLFROQAGSGLSEBDAPEREGILAYVMSSENFSPRGPPSSSSISSTSPPSYDSV	1980
QY	1980	TRATSDNLOVAGSDYSHSEDLADFPSPDRDRESIV	2015
Db	1981	TRATSDNLOVAGSDYSHSEDLADFPSPDRDRESIV	2016
RESULT 17			
AAB82240			
ID	AAB82240	standard; protein; 2016 AA.	
XX	XX		
AC	AAB82240;		
XX	XX		
DT	21-JUN-2001 (first entry)		
XX	XX		
DE	Human SCNSA mutant D114N.		
XX	XX		
KM	SCNSA; long QT syndrome; LQTS; cardiovascular disease;		
KW	Romano-Ward syndrome; diagnosis; prognosis; therapy; drug screening;		
KW	mutant; mutant.		
XX	XX		
OS	Homo sapiens.		
XX	XX		
PN	WO200124681-A2.		
XX	XX		
PD	12-APR-2001.		
XX	XX		
PF	09-AUG-2000; 2000WO-US021660.		
XX	XX		
PR	09-AUG-1999; 99US-0147488P.		
XX	XX		
PR	17-MAR-2000; 2000US-0190057P.		
XX	XX		
PA	(UTAH) UNIV UTAH RES FOUND.		
XX	XX		
PI	Keating MT, Splawski I;		
XX	XX		
DR	WPI; 290564/30.		
XX	XX		
PT	New KVLQT1 and SCNSA genes, which contains alterations or mutations,		
PT	useful in diagnostic/prognostic or drug screening methods, particularly in		
PT	mutational analyses for screening individuals with or at risk for long QT		
XX	syndrome.		
XX	XX		

QY 1860 VLGESEMDALKIOMEKEMANPSKISYEPTITTLRRKHEVSAMVIOARFRRLQRS 1919
 DB 1861 VLGESEMDALKIOMEKEMANPSKISYEPTITTLRRKHEVSAMVIOARFRRLQRS 1920
 QY 1920 LKHASFLFQOAGSGLSEEDAPEREGLIAYVMSSESRPLGPPSSSSISSTSPPPSYDSV 1979
 DB 1921 LKHASFLFQOAGSGLSEEDAPEREGLIAYVMSSESRPLGPPSSSSISSTSPPPSYDSV 1980
 QY 1980 TRATSDNLQVRGSDYSHSEDLADFPSPPRDRESIV 2015
 DB 1981 TRATSDNLQVRGSDYSHSEDLADFPSPPRDRESIV 2016

RESULT 18
 AAB82243
 ID AAB82243 standard; protein; 2016 AA.
 XX
 AC AAB82243;
 XX
 DT 21-JUN-2001 (first entry)
 XX
 DE Human SCN5A mutant R1623L.
 XX
 KW SCN5A; long QT syndrome; LQTS; cardiovascular disease;
 KW Romano-Ward syndrome; diagnosis; prognosis; therapy; drug screening;
 KW mutant; mutcin.
 XX
 OS Homo sapiens.
 XX
 PN WO200124681-A2.
 XX
 PD 12-APR-2001.
 XX
 PF 09-AUG-2000; 2000WO-US021660.
 XX
 PR 09-AUG-1999; 99US-0147488P.
 PR 17-MAR-2000; 2000US-0190057P.
 XX
 PA (UTAH) UNITV UTAH RES FOUND.
 XX
 PI Keating MT, Splawski I;
 XX
 DR WPI; 2001-290564/30.
 XX
 PT New KVLQT1 and SCN5A genes, which contains alterations or mutations,
 PT useful in diagnostic/prognostic or drug screening methods, particularly in
 PT mutational analyses for screening individuals with or at risk for long QT
 PT syndrome.
 XX
 PS Claim 31; Page; 76pp; English.
 XX
 CC The present sequence is that of the claimed R1623T mutant of the human
 CC SCN5A protein. The mutant is encoded by an SCN5A mutant gene in which a
 CC G/T mutation alters codon 1623 from CGA to CTA. Mutations of the SCN5A
 CC gene are implicated in Romano-Ward syndrome, the autosomal dominant form
 CC of long QT syndrome (LQTS). Mutations newly discovered in the SCN5A gene
 CC lead to the following amino acid alterations in the encoded protein:
 CC D1114N, L1501V, delP1617, R1623L, E1784K and S1787N. Knowledge of the
 CC mutations provides means for assessing a risk in a human subject for
 CC LQTS, for diagnosing a mutation which causes LQTS, and for screening for
 CC drugs useful in treating a mutation which causes LQTS in the SCN5A gene.
 CC Note: The present sequence is not shown in the specification but is
 CC derived from the KVLQT-1 sequence given in the Sequence Listing (see
 CC AAB82220)
 XX
 SQ Sequence 2016 AA;

Query Match 99.4%; Score 10425.5; DB 4; Length 2016;
 Best Local Similarity 99.5%; Pred. No. 0;
 Matches 2005; Conservative 2; Mismatches 8; Indels 1; Gaps 1;

QY 1 MANFLPRTGTSFRFRFTRESLAIKRMMAEKQARGSTTLQESREGLPREEAPRPOLDQA 60
 |||

DB 1 MANFLPRTGTSFRFRFTRESLAIKRMMAEKQARGSTTLQESREGLPREEAPRPOLDQA 60
 QY 61 SKKLDELIGNPPQELIGLELEDLPFYSGQTKFYLNKTKTFRPSATNALVLSFPHI 120
 DB 61 SKKLDELIGNPPQELIGLELEDLPFYSGQTKFYLNKTKTFRPSATNALVLSFPHV 120
 QY 121 RRAAVKILVHSLFNNLIMCTIITNCFMAQHPDPPTKVEYTFPAIYTFESLVKILARG 180
 DB 121 RRAAVKILVHSLFNNLIMCTIITNCFMAQHPDPPTKVEYTFPAIYTFESLVKILAA 180
 QY 181 FCLAAFTFLRDEPMNLDFSVIIIMAYTTEFVDIGANVSALRTFVRLAKTISVISGLKTI 240
 DB 181 FCLAAFTFLRDEPMNLDFSVIIIMAYTTEFVDIGANVSALRTFVRLAKTISVISGLKTI 240
 QY 241 GALIOSVKKLADVNTLVTPCLSVFPLIGLQFMGNLRHKCVNFPALNKTNSVEADGIV 300
 DB 241 GALIOSVKKLADVNTLVTPCLSVFPLIGLQFMGNLRHKCVNFPALNKTNSVEADGIV 300
 QY 301 WESLDLYSDPENNYLLKNGTSDVLLCGNSDAGTCPEGRCCLKAGENPDHGYTSFDSFAM 360
 DB 301 WESLDLYSDPENNYLLKNGTSDVLLCGNSDAGTCPEGRCCLKAGENPDHGYTSFDSFAM 360
 QY 361 AFLALFRLMTQDCWERYLQOTLRSAKITYMIFPMLVIFLGSFYLVNLLAVVAMAYEON 420
 DB 361 AFLALFRLMTQDCWERYLQOTLRSAKITYMIFPMLVIFLGSFYLVNLLAVVAMAYEON 420
 QY 421 QATIAETEKEKRFQEAEMELKKEHEALTIRGVTVSSLSLEMSPLAPVNSHERSKRK 480
 DB 421 QATIAETEKEKRFQEAEMELKKEHEALTIRGVTVSSLSLEMSPLAPVNSHERSKRK 480
 QY 481 RWSGTEGCRDLRPKSDSDGPRAMNHLSTRGLSRTSMKRSRSGSTFTRRRDLGSE 540
 DB 481 RWSGTEGCRDLRPKSDSDGPRAMNHLSTRGLSRTSMKRSRSGSTFTRRRDLGSE 540
 QY 541 ADFADDENSTAGESESHRTSLVPPLRRTSAQCPSPGTSAPGHALGKNSYDNCV 600
 DB 541 ADFADDENSTAGESESHRTSLVPPLRRTSAQCPSPGTSAPGHALGKNSYDNCV 600
 QY 601 VSLGAGDPEATSPGSHLLRPVMLEHPDPTTSPSEEGPQMLTISOACVGFEEBPGRQ 660
 DB 601 VSLGAGDPEATSPGSHLLRPVMLEHPDPTTSPSEEGPQMLTISOACVGFEEBPGRQ 660
 QY 661 RALSASVLTALAELEESRRHKCPCMRLAQRVYIWCPCPLMMSIKGVKLVMWDPFD 720
 DB 661 RALSASVLTALAELEESRRHKCPCMRLAQRVYIWCPCPLMMSIKGVKLVMWDPFD 720
 QY 721 LTTTCIYVNLTFMALBHYNNTSEFEEMLYQGNLVFTGIFTAEMTFKIIADPPYYPQ 780
 DB 721 LTTTCIYVNLTFMALBHYNNTSEFEEMLYQGNLVFTGIFTAEMTFKIIADPPYYPQ 780
 QY 781 WNIFDSIIVILSLMELGSRMSNLSVLRSPRLRVFKLAKSWPTLNTLIIKIIINSVGLG 840
 DB 781 WNIFDSIIVILSLMELGSRMSNLSVLRSPRLRVFKLAKSWPTLNTLIIKIIINSVGLG 840
 QY 841 NLTVLATIIIVIPAVNGQLGKXNSLRSDSGLLPWHMMDPFAHLIIFRILCGEIT 900
 DB 841 NLTVLATIIIVIPAVNGQLGKXNSLRSDSGLLPWHMMDPFAHLIIFRILCGEIT 900
 QY 901 ETTMWDCEVSQSCLVFLVLMVIGNLVNLFALILSSFSADNLTAPDEBREMNLQ 960
 DB 901 ETTMWDCEVSQSCLVFLVLMVIGNLVNLFALILSSFSADNLTAPDEBREMNLQ 960
 QY 961 LALARIQGLRFVKKRTWDFCCGLLRQRPQKPAALAAQGLPSCITATYSPPEPTEKYP 1020
 DB 961 LALARIQGLRFVKKRTWDFCCGLLRQRPQKPAALAAQGLPSCITATYSPPEPTEKYP 1020
 QY 1021 PTRKREBEGEOPGCGRPDPBPVCVPIAAESDTPDOEBDENSTGTEBESSK-QDSQ 1079
 DB 1021 PTRKREBEGEOPGCGRPDPBPVCVPIAAESDTPDOEBDENSTGTEBESSK-QDSQ 1080
 QY 1080 PVSGGPEAPDPDSRTWSQVATASSEABASASQADWRQWKAEPQAPCGEFTPEDSCSGS 1139
 DB 1081 PVSGWPRPDPDSRTWSQVATASSEABASASQADWRQWKAEPQAPCGEFTPEDSCSGS 1140

QY	1140	TADMTTAAELLEQIPDLGDGVKPEDCFTEGCVRRGCCCAVDTTQAGKYMNLAKTCYH	1199
Db	1141	TADMTTAAELLEQIPDLGDGVKPEDCFTEGCVRRGCCCAVDTTQAGKYMNLAKTCYH	1200
QY	1200	IVESMFETFEFIFMILLSSGALAEEDLYLEERKTIKYLEEADQMFFYYVLEMLAKMVA	1259
Db	1201	IVESMFETFEFIFMILLSSGALAEEDLYLEERKTIKYLEEADQMFFYYVLEMLAKMVA	1260
QY	1260	YGFKKYFTNACWMLDPLIVDVSLVSLVANTLGPALMEGPIKSLRTLRLRLPALSRREGM	1319
Db	1261	YGFKKYFTNACWMLDPLIVDVSLVSLVANTLGPALMEGPIKSLRTLRLPALSRREGM	1320
QY	1320	RYVYNALVGALPSIMNVLVCLFVWLFSIMGNVLFGKRGRCINQTEGDPMLNYTIVN	1379
Db	1321	RYVYNALVGALPSIMNVLVCLFVWLFSIMGNVLFGKRGRCINQTEGDPMLNYTIVN	1380
QY	1380	KSQCESINTLNGELVMTKYKVPFDVAGVYALALQVAFPKGMDIMYAAVDSRGYEOPQW	1439
Db	1381	KSQCESINTLNGELVMTKYKVPFDVAGVYALALQVAFPKGMDIMYAAVDSRGYEOPQW	1440
QY	1440	EYNLYMTIYEVYFTIIFGSGFFTLNLFIGYIINENQOKKLGQODIEMTEBOKKTYNMMKK	1499
Db	1441	EYNLYMTIYEVYFTIIFGSGFFTLNLFIGYIINENQOKKLGQODIEMTEBOKKTYNMMKK	1500
QY	1500	LGSKKPKQPIPRPLANKQGFEDLVYTKQAPDVTLMPIICLNMVMYMETDQSEKINIL	1559
Db	1501	LGSKKPKQPIPRPLANKQGFEDLVYTKQAPDVTLMPIICLNMVMYMETDQSEKINIL	1560
QY	1560	AKINILFVALFTGBCIVGLAALRRHYFTNSWNIPEFVVVILISIVGLSDIIOKTFPSPT	1619
Db	1561	AKINILFVALFTGBCIVGLAALRRHYFTNSWNIPEFVVVILISIVGLSDIIOKTFPSPT	1620
QY	1620	LFRTYIRLARIGRIIRLRIRGAGGIRTLPALMMSLPALPNIGLLFLVMFTYSIRGMANPA	1679
Db	1621	LFRTYIRLARIGRIIRLRIRGAGGIRTLPALMMSLPALFNIGLLFLVMFTYSIRGMANPA	1680
QY	1680	YVKKMEAGIDDMENFQTFANSMCLCFQRTSAGMGGLSPILANTGPYCDPTLPNSNGSRG	1739
Db	1681	YVKKMEAGIDDMENFQTFANSMCLCFQRTSAGMGGLSPILANTGPYCDPTLPNSNGSRG	1740
QY	1740	DCGSPAVGILFETTYIIISPLIVNMVTAIILLENFSVATEESTEPLESDDFMFEIWEK	1799
Db	1741	DCGSPAVGILFETTYIIISPLIVNMVTAIILLENFSVATEESTEPLESDDFMFEIWEK	1800
QY	1800	FDPERQTOITEYSVLSDPADLASEPLRIAKPNQOISLINMDLPMVSGDRHCHMDILPAFTKR	1859
Db	1801	FDPERQTOITEYSVLSDPADLASEPLRIAKPNQOISLINMDLPMVSGDRHCHMDILPAFTKR	1860
QY	1860	VLGESGEMDALKIQWEEKFMANPESKISYEPIITTLTRKHEEVSAMVIOQAFRRHLQORS	1919
Db	1861	VLGESGEMDALKIQWEEKFMANPESKISYEPIITTLTRKHEEVSAMVIOQAFRRHLQORS	1920
QY	1920	LKHASFILRQOAGSLSEDAPEERBGLIAYVNSENFSRPLCPSPSSSISSTSPSPSYDSV	1979
Db	1921	LKHASFILRQOAGSLSEDAPEERBGLIAYVNSENFSRPLCPSPSSSISSTSPSPSYDSV	1980
QY	1980	TRATSDNLOVNGSDYSHSEDLADPPSPDPRORESTIV	2015
Db	1981	TRATSDNLOVNGSDYSHSEDLADPPSPDPRORESTIV	2016
RESULT 19			
AAW23994			
ID	AAW23994	standard; protein; 2016 AA.	
XX	AAW23994;		
XX	AC		
XX	DT	06-JUL-1998 (first entry)	
XX	DE	Human hH1 sodium channel protein.	
XX	KW	Ion channel; sodium channel; hH1; human; cardiac cell; heart; pacemaker;	

KM	gene therapy.
XX	
OS	Homo sapiens.
XX	
PN	WO9802040-A1.
XX	
PD	22-JAN-1998.
XX	
PP	04-APR-1997; 97WO-US005556.
XX	
PR	17-JUL-1996; 96US-00682433.
XX	
PA	(MEDT) MEDTRONIC INC.
XX	
PI	Stokes KB, Morrisette J;
XX	
DR	WPI; 1998-110247/10.
XX	
DR	N-PSDB; AAV09029.
PT	System for delivering genetic material to heart - comprises reservoir,
PT	catheter and optionally pacing electrode for delivering ion-channel
XX	protein, useful for, e.g. improving sensing by pacemaker.
XX	
PS	Disclosure; Page 41-47; 73pp; English.

Disclosure; Page 41-47; 73pp; English.

This protein comprises the human hH1 voltage-regulated sodium channel protein that can be used in a novel system for enhancing cardiac signal sensing by cardiac pacemakers through genetic treatment. A claimed system for delivering genetic material (GM) comprises a reservoir containing GM and a device for delivering it to myocardial cells (MC) at a specific location. The GM increases the amplitude of the cardiac signal, improving the signal-to-noise (S/N) ratio that is sensed by the electrode of a pacemaker. Also claimed are: (1) an implantable delivery system comprising a reservoir for GM which increases the expression of ion channels in MC and system for delivering this through a catheter, the tip of which engages MC at the chosen location, and (2) a system similar to (1) comprising a pacing electrode on an inner wall of the heart, close to the site where the GM is delivered. The system is used for delivery of an ion-channel GM which causes depolarisation of atrial and ventricular MC and improves the sensing of cardiac signals by the pacemaker and the S/N ratio of atrial P-waves. The preferred GM comprises DNA (see AAV09029) or RNA encoding hH1.

Sequence 2016 AA; SQ

Query Match	99.4%	Score 10421.5;	DB 2;	Length 2016;
Best Local Similarity	99.4%;	Pred. No. 0;		
Matches 2004; Conservative	3;	Mismatches	8;	Indels 1; Gaps 1;

QY	1	MANFLTPGTSFRPFTESLAALEKNAEQAGSTLIQSRGLPEEAPRQLOQA	60
QY	1	MANFLTPGTSFRPFTESLAALEKNAEQAGSTLIQSRGLPEEAPRQLOQA	60
Db	1	MANFLTPGTSFRPFTESLAALEKNAEQAGSTLIQSRGLPEEAPRQLOQA	60
QY	61	SKLPLDLYGNPPOELIGHPLEDDLPFYSQTKTFIVLNGKTIIPRSATNALYVSPFPI	120
Db	61	SKLPLDLYGNPPOELIGHPLEDDLPFYSQTKTFIVLNGKTIIPRSATNALYVSPFPI	120
QY	121	RRAAKILVHSLFNNLIMCTILTNCFVMAQDPPMTKYEVETAIITFESLVKILARG	180
Db	121	RRAAKILVHSLFNNLIMCTILTNCFVMAQDPPMTKYEVETAIITFESLVKILARA	180
QY	181	FCLHAFTEFLRDPNMWLDPSVIIMATTFEVDLGNVSALIRTRVLRALKTISVIGLKTIIV	240
Db	181	FCLHAFTEFLRDPNMWLDPSVIIMATTFEVDLGNVSALIRTRVLRALKTISVIGLKTIIV	240
QY	241	GALIOSVKKLADVWMLITVFCLSVFPALIGLOFMENLRKCYRNFTALNGTGSVEADGLV	300
Db	241	GALIOSVKKLADVWMLITVFCLSVFPALIGLOFMENLRKCYRNFTALNGTGSVEADGLV	300
QY	301	WESIDLVLSDPENYLLKNGTSDVLLCGNSSDAGTCPEBGRCLKAGENDHGTSTPDSFAM	360
Db	301	WESIDLVLSDPENYLLKNGTSDVLLCGNSSDAGTCPEBGRCLKAGENDHGTSTPDSFAM	360


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QY 361 AFALFRLMTOPCMBELYOQTLRSAGKIYMIFFMLVIFGSPYLVNLLAVAMAYEON 420
DB 361 AFALFRLMTOPCMBELYOQTLRSAGKIYMIFFMLVIFGSPYLVNLLAVAMAYEON 420
QY 421 QATIAETEKEKRRFOEAMEMLKKEHEALTIRGVTVSRSSLEMSPLAPVNSHERSKRK 480
DB 421 QATIAETEKEKRRFOEAMEMLKKEHEALTIRGVTVSRSSLEMSPLAPVNSHERSKRK 480
QY 481 RMSGTEBEGGERLPKSDSEDDPRAMNHLSTRGLSRITSMKPRSSRGSIPTRRRDDLSE 540
DB 481 RMSGTEBEGGERLPKSDSEDDPRAMNHLSTRGLSRITSMKPRSSRGSIPTRRRDDLSE 540
QY 541 ADFADENSTAGESHRTSLVPWPLRRTSAQOGSPGTSAPGHALHKKSTYDCNGV 600
DB 541 ADFADENSTAGESHRTSLVPWPLRRTSAQOGSPGTSAPGHALHKKSTYDCNGV 600
QY 601 VSLGAGDEEATSPGSHLARPVMLEHPDPTTTPSEBPGPOMLTQAPCVDGFEPEGARQ 660
DB 601 VSLGAGDEEATSPGSHLARPVMLEHPDPTTTPSEBPGPOMLTQAPCVDGFEPEGARQ 660
QY 661 RALSAVSUTLSALELEBSRHKCPCCMNFLAORYLIMECCPLMSTKQCVKLVNMDPTD 720
DB 661 RALSAVSUTLSALELEBSRHKCPCCMNFLAORYLIMECCPLMSTKQCVKLVNMDPTD 720
QY 721 LITMCIYVANTLFMALHEHNMTSEPEMLQVGNLVPFGITFAEMTFKIIALDPYYPQOG 780
DB 721 LITMCIYVANTLFMALHEHNMTSEPEMLQVGNLVPFGITFAEMTFKIIALDPYYPQOG 780
QY 781 WNIPIISIIYLSIMEGLSRMSNLVLSRFLRLRVFKLAKSWPTNTLIIKIIIGNSVGALG 840
DB 781 WNIPIISIIYLSIMEGLSRMSNLVLSRFLRLRVFKLAKSWPTNTLIIKIIIGNSVGALG 840
QY 841 NITLVLAITVIFPAVYGMQLFGKNTSELDSGSLPRWMMDFPAFLITRIICGEVI 900
DB 841 NITLVLAITVIFPAVYGMQLFGKNTSELDSGSLPRWMMDFPAFLITRIICGEVI 900
QY 901 ETMMDCMEVSGSLCLVFLVAVIGNLVNLFALILSSFSANLTPADERMANNIQ 960
DB 901 ETMMDCMEVSGSLCLVFLVAVIGNLVNLFALILSSFSANLTPADERMANNIQ 960
QY 961 LALARIORGARFVKRTTDFCCGLRORPQKPAALAAQOLPSCATPSPPEPEKVP 1020
DB 961 LALARIORGARFVKRTTDFCCGLRORPQKPAALAAQOLPSCATPSPPEPEKVP 1020
QY 1021 PTRKETREBEGEPQCGTIPGDEPVCVPIAVESTDOEEDENS LGTEBESSK-QESQ 1079
DB 1021 PTRKETREBEGEPQCGTIPGDEPVCVPIAVESTDOEEDENS LGTEBESSK-QESQ 1079
QY 1080 PVSQGPAPDPSRTWSQVATASSBAEASASQADWRQOKAPQAPGCGETPEDSCSBS 1139
DB 1080 PVSQGPAPDPSRTWSQVATASSBAEASASQADWRQOKAPQAPGCGETPEDSCSBS 1139
QY 1140 TADMNTNTELEQIDPLGDVADPCFTEGCVRRCPCCAVDTTAPAGKVMRLKTCYH 1199
DB 1140 TADMNTNTELEQIDPLGDVADPCFTEGCVRRCPCCAVDTTAPAGKVMRLKTCYH 1199
QY 1141 TADMNTNTELEQIDPLGDVADPCFTEGCVRRCPCCAVDTTAPAGKVMRLKTCYH 1200
DB 1141 TADMNTNTELEQIDPLGDVADPCFTEGCVRRCPCCAVDTTAPAGKVMRLKTCYH 1200
QY 1200 IVESHMPEFTIIFMLISSGALAFEDIIYLBERTIKVILEVADKMFYVFLVLEMLLKVA 1259
DB 1200 IVESHMPEFTIIFMLISSGALAFEDIIYLBERTIKVILEVADKMFYVFLVLEMLLKVA 1259
QY 1260 YGFKKYFTNANCMWDLFLVDVSLVAVNTLGFAMGPKSLRTLRALRPLALSRFGM 1319
DB 1260 YGFKKYFTNANCMWDLFLVDVSLVAVNTLGFAMGPKSLRTLRALRPLALSRFGM 1319
QY 1320 YGFKKYFTNANCMWDLFLVDVSLVAVNTLGFAMGPKSLRTLRALRPLALSRFGM 1320
DB 1320 YGFKKYFTNANCMWDLFLVDVSLVAVNTLGFAMGPKSLRTLRALRPLALSRFGM 1320
QY 1330 RVVVALVGAIPISINAVLLVCLIFMLIFSIMGVNFAGKFGCINOTEGDPLANTYNN 1379
DB 1330 RVVVALVGAIPISINAVLLVCLIFMLIFSIMGVNFAGKFGCINOTEGDPLANTYNN 1379
QY 1380 KSQCESLNTLGBELVNTKRVNFDNVAGYLLALLQVATFGKMDIMYAAVDSRGYEQPOM 1439
DB 1380 KSQCESLNTLGBELVNTKRVNFDNVAGYLLALLQVATFGKMDIMYAAVDSRGYEQPOM 1439
QY 1440 EVNLVMTYIFVFIIFGSPFTLNLRTIGVLIIDFNQKKKGGQDIFMTBEQKKYNNAMKX 1499
DB 1440 EVNLVMTYIFVFIIFGSPFTLNLRTIGVLIIDFNQKKKGGQDIFMTBEQKKYNNAMKX 1499

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DB 1441 EVNLVMTYIFVFIIFGSPFTLNLRTIGVLIIDFNQKKKGGQDIFMTBEQKKYNNAMKX 1500
QY 1500 LGSKKQKPIPRPLANKYGFIFDIYTKQAFVYITMFLCLMNTVMVETDDOSPKNINL 1559
DB 1501 LGSKKQKPIPRPLANKYGFIFDIYTKQAFVYITMFLCLMNTVMVETDDOSPKNINL 1560
QY 1560 AKINLFAVITGECTVLAALRHYPFTNSNNIPFVVVVISYGVVLSDIIOKFFSPT 1619
DB 1561 AKINLFAVITGECTVLAALRHYPFTNSNNIPFVVVVISYGVVLSDIIOKFFSPT 1620
QY 1620 LFRVIRLARIGRIILIRINGANGIRTLFALMMSIPALFNIGILLFLVNFYISIPGMANPA 1679
DB 1621 LFRVIRLARIGRIILIRINGANGIRTLFALMMSIPALFNIGILLFLVNFYISIPGMANPA 1680
QY 1680 YVKEWAGIDDMFNFTFANSMCLFQITTSAGMDGLSPIIANTGPYCDPTLPNSNGSRG 1739
DB 1681 YVKEWAGIDDMFNFTFANSMCLFQITTSAGMDGLSPIIANTGPYCDPTLPNSNGSRG 1740
QY 1740 DCGSPAVGILFPTTYIIISPLIVNMYTAIILENFSVATESTEPLSEDDPFMEIEMK 1799
DB 1741 DCGSPAVGILFPTTYIIISPLIVNMYTAIILENFSVATESTEPLSEDDPFMEIEMK 1800
QY 1800 FDPPEATQFIEYSVLSDFADALSEPRLAKPNQISLIINDLPMVSGDRTHCWDILFAFTKR 1859
DB 1801 FDPPEATQFIEYSVLSDFADALSEPRLAKPNQISLIINDLPMVSGDRTHCWDILFAFTKR 1860
QY 1860 VLGSEGDMDALKIOMEKFMANPSKISYEPITTTLRKHEEVSAMVIORAFRRHLQRS 1919
DB 1861 VLGSEGDMDALKIOMEKFMANPSKISYEPITTTLRKHEEVSAMVIORAFRRHLQRS 1920
QY 1920 LKHASFILROQAGSGLSEDEAPEREGLIAYVMSNFSPDLPPSSSISSTSPSPSYSV 1979
DB 1921 LKHASFILROQAGSGLSEDEAPEREGLIAYVMSNFSPDLPPSSSISSTSPSPSYSV 1980
QY 1980a TRATSNDNLQVRSYSHSEDLADPPSPDRRESIV 2015
DB 1981 TRATSNDNLQVRSYSHSEDLADPPSPDRRESIV 2016

RESULT 20
AAB82242
ID AAB82242 standard; protein, 2015 AA.
XX
XX AAB82242;
AC
XX
XX 21-JUN-2001 (first entry)
DT
XX
XX
DE Human SCNSA mutant delP1617.
XX
XX SCNSA; Long QT syndrome; LQTS; cardiovascular disease;
KM Romano-Ward syndrome; diagnosis; prognosis; therapy; drug screening;
KW mutant; mutcin.
XX
OS Homo sapiens.
XX
XX W0200124681-A2.
XX
XX 12-APR-2001.
XX
XX 09-AUG-2000; 2000WO-US021660.
XX
XX 09-AUG-1999; 99US-0147488P.
XX
XX 17-MAR-2000; 2000US-0190057P.
XX
XX (UTAH ) UNIV UTAH RBS FOUND.
XX
XX Keating MT, Splawski I;
XX
XX WPI; 2001-290564/30.
XX
XX New KVLQTI and SCNSA genes, which contains alterations or mutations,
XX useful in diagnostic/prognostic or drug screening methods, particularly in
XX
XX

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PT mutational analyses for screening individuals with or at risk for long QT syndrome.

Claim 31; Page: 76pp; English.

CC The present sequence is that of the claimed delP617 mutant of the human
CC SCN5A protein. The mutant is encoded by an SCN5A mutant gene carrying a
CC deletion of the TTC codon for Phe-1617. Mutations of the SCN5A gene are
CC implicated in Romano-Ward syndrome, the autosomal dominant form of Long
CC QT syndrome (LQTS). Mutations newly discovered in the SCN5A gene lead to
CC the following amino acid alterations in the encoded protein: D1114N,
CC L1501V, delP617, R1623I, E1764K and S1787N. Knowledge of the mutations
CC provides means for assessing a risk in a human subject for LQTS, for
CC diagnosing a mutation which causes LQTS, and for screening for drugs
CC useful in treating a human having a mutation in the SCN5A gene. Note: The
CC present sequence is not shown in the specification but is derived from
CC the KIVQT-1 sequence given in the Sequence Listing (see AAB82220)

XX Sequence 2015 AA;

Query Match 99.3%; Score 10416; DB 4; Length 2015;

Best Local Similarity 99.5%; Pred. No. 0;

Matches 2005; Conservative 2; Mismatches 7; Indels 2; Gaps 2;

QY 1 MANFLIPRGTSFRRFTRSLAIEKMAEKQARGSTTLQESREGLPREEAPRPQLDQA 60
DB 1 MANFLIPRGTSFRRFTRSLAIEKMAEKQARGSTTLQESREGLPREEAPRPQLDQA 60
QY 61 SKKLDELGNPQOELIGEPLEDLPFYSTQKTFIYLNKQKTIFFRSATNALVYLSPPFI 120
DB 61 SKKLDELGNPQOELIGEPLEDLPFYSTQKTFIYLNKQKTIFFRSATNALVYLSPPFI 120
QY 121 RBAAYKIIIVHSLENNLIMCTIITNCVMAQHDPPMTKXVEYTFATYTESVYLARG 180
DB 121 RBAAYKIIIVHSLENNLIMCTIITNCVMAQHDPPMTKXVEYTFATYTESVYLARG 180
QY 181 PCLHAFPLRDPWNLDSVITIMATTEFVDIGNSALRTPRVLRALKTISVLSGLKTIY 240
DB 181 PCLHAFPLRDPWNLDSVITIMATTEFVDIGNSALRTPRVLRALKTISVLSGLKTIY 240
QY 181 PCLHAFPLRDPWNLDSVITIMATTEFVDIGNSALRTPRVLRALKTISVLSGLKTIY 240
DB 181 PCLHAFPLRDPWNLDSVITIMATTEFVDIGNSALRTPRVLRALKTISVLSGLKTIY 240
QY 241 GALLISVKGLADVMVLTVCLSVFALIGLQFMGNLRHKCVANFTALNGTNGSVEADGIV 300
DB 241 GALLISVKGLADVMVLTVCLSVFALIGLQFMGNLRHKCVANFTALNGTNGSVEADGIV 300
QY 241 GALLISVKGLADVMVLTVCLSVFALIGLQFMGNLRHKCVANFTALNGTNGSVEADGIV 300
DB 241 GALLISVKGLADVMVLTVCLSVFALIGLQFMGNLRHKCVANFTALNGTNGSVEADGIV 300
QY 301 WESLDLYLSDPENYLLKNGTSDVLLCGNSSDAGTCEGYRCLKAGBNPDHGYTSPDFAM 360
DB 301 WESLDLYLSDPENYLLKNGTSDVLLCGNSSDAGTCEGYRCLKAGBNPDHGYTSPDFAM 360
QY 301 WESLDLYLSDPENYLLKNGTSDVLLCGNSSDAGTCEGYRCLKAGBNPDHGYTSPDFAM 360
DB 301 WESLDLYLSDPENYLLKNGTSDVLLCGNSSDAGTCEGYRCLKAGBNPDHGYTSPDFAM 360
QY 361 AFLALFRLMTODCWEELYYOQTLRSAGKIYMFMLVIFLGSFYLVNLLAVAMAYEEN 420
DB 361 AFLALFRLMTODCWEELYYOQTLRSAGKIYMFMLVIFLGSFYLVNLLAVAMAYEEN 420
QY 421 QATIAETEERKRFQAMEMLKKEHEALITRGVDYTSRSLSEMSPLAPVNSHERSKRK 480
DB 421 QATIAETEERKRFQAMEMLKKEHEALITRGVDYTSRSLSEMSPLAPVNSHERSKRK 480
QY 481 RMSSGTECEGDELRPKSDEDEGRAMNHLSTLRGLSRTSMKPRSRSRGSIFTRRDLSGE 540
DB 481 RMSSGTECEGDELRPKSDEDEGRAMNHLSTLRGLSRTSMKPRSRSRGSIFTRRDLSGE 540
QY 481 RMSSGTECEGDELRPKSDEDEGRAMNHLSTLRGLSRTSMKPRSRSRGSIFTRRDLSGE 540
DB 481 RMSSGTECEGDELRPKSDEDEGRAMNHLSTLRGLSRTSMKPRSRSRGSIFTRRDLSGE 540
QY 541 ADPADENSTAGSESEHRTSLVPMPLRRTSAQGOSPGTSAFGALHKKNSYDNCNV 600
DB 541 ADPADENSTAGSESEHRTSLVPMPLRRTSAQGOSPGTSAFGALHKKNSYDNCNV 600
QY 541 ADPADENSTAGSESEHRTSLVPMPLRRTSAQGOSPGTSAFGALHKKNSYDNCNV 600
DB 541 ADPADENSTAGSESEHRTSLVPMPLRRTSAQGOSPGTSAFGALHKKNSYDNCNV 600
QY 601 VSLILGADPEATSPGSHLRPVMLLEHPPTTTPSEEPGPOMLTSAQPCVDGFEERGAQ 660
DB 601 VSLILGADPEATSPGSHLRPVMLLEHPPTTTPSEEPGPOMLTSAQPCVDGFEERGAQ 660
QY 661 RALSAYSVLTSALBELLESRRHKPCPCMNRLAORYLLIWECCLPMWSIKQGVKLVMNDPTD 720
DB 661 RALSAYSVLTSALBELLESRRHKPCPCMNRLAORYLLIWECCLPMWSIKQGVKLVMNDPTD 720
QY 721 LITTMCIIVNTLPMALHNTMTSEFEBMLQVGNLVFTGIFTAEMTKITIALDPYFFQOG 780

DB 721 LITTMCIIVNTLPMALHNTMTSEFEBMLQVGNLVFTGIFTAEMTKITIALDPYFFQOG 780
QY 781 WNIIPDSIIVLISLMEJGLSMNSULSVRSRLLRVKLAWSPTLNTLTKITONSVALG 840
DB 781 WNIIPDSIIVLISLMEJGLSMNSULSVRSRLLRVKLAWSPTLNTLTKITONSVALG 840
QY 841 NLTLVATITVFYAVVGMOLFGKNYSRLRSDSGLPRMHMDPFNAFLIIFRILCGEWI 900
DB 841 NLTLVATITVFYAVVGMOLFGKNYSRLRSDSGLPRMHMDPFNAFLIIFRILCGEWI 900
QY 901 ETMMDCMVEVSQSICLLVFLVWVIGNLVYVNLFLALLSSFSADNLTAPDERENMTQ 960
DB 901 ETMMDCMVEVSQSICLLVFLVWVIGNLVYVNLFLALLSSFSADNLTAPDERENMTQ 960
QY 961 LALARIQGRIFVKRTTWDCCLLROKRPQKPAALAAQGLPBCIATPYSPPEPEKVP 1020
DB 961 LALARIQGRIFVKRTTWDCCLLROKRPQKPAALAAQGLPBCIATPYSPPEPEKVP 1020
QY 1021 PTRKETRFEBGEOPGQTPDPPVPCVPIVAASDTPDDEBENSIGTEESSK-OESQ 1079
DB 1021 PTRKETRFEBGEOPGQTPDPPVPCVPIVAASDTPDDEBENSIGTEESSK-OESQ 1079
QY 1080 PVSGCPAPAPDSRTWSQVSATASSEASASQADWRQWKAEPQAPCGETPEDSCSEG 1139
DB 1080 PVSGCPAPAPDSRTWSQVSATASSEASASQADWRQWKAEPQAPCGETPEDSCSEG 1139
QY 1140 TADMTNTAELLBOQIPDVGQVQKDECFTEGCRCRCCCAVDTTQAGKXWMLRKTQYH 1199
DB 1140 TADMTNTAELLBOQIPDVGQVQKDECFTEGCRCRCCCAVDTTQAGKXWMLRKTQYH 1199
QY 1200 IVEHSWFETPIIFMILSSGALAFEDYLERKTIKYLEYADMFTYVFLBMLAKVA 1259
DB 1200 IVEHSWFETPIIFMILSSGALAFEDYLERKTIKYLEYADMFTYVFLBMLAKVA 1259
QY 1260 YGFKKYFTNACWLDPLIVDVSLVSVANTLGFANMGPISLRTLRALRPLRALSREEM 1319
DB 1260 YGFKKYFTNACWLDPLIVDVSLVSVANTLGFANMGPISLRTLRALRPLRALSREEM 1319
QY 1320 RYVVALVGAIPSTIMNVLYVCLIFWLIIFSINGVLFNGKRGRCINOTEGDPLNYTTVNN 1379
DB 1320 RYVVALVGAIPSTIMNVLYVCLIFWLIIFSINGVLFNGKRGRCINOTEGDPLNYTTVNN 1379
QY 1380 KSGCESLNTGELVMTYKVNFDVNGVGYALLOVAFKGMMDIMYAAVDSRGEOPQW 1439
DB 1380 KSGCESLNTGELVMTYKVNFDVNGVGYALLOVAFKGMMDIMYAAVDSRGEOPQW 1439
QY 1440 EYNLYMYIYFVIFIFGSPFTLNLFIGVIIDNFNOQKKLGGQDIEMTEBOKKYNNAMKK 1499
DB 1440 EYNLYMYIYFVIFIFGSPFTLNLFIGVIIDNFNOQKKLGGQDIEMTEBOKKYNNAMKK 1499
QY 1500 LGSKKPOKPIPRPLNKYQGFIDVLYQKAPVYTIMPLICLNMNTMNETDQSEKINIL 1559
DB 1500 LGSKKPOKPIPRPLNKYQGFIDVLYQKAPVYTIMPLICLNMNTMNETDQSEKINIL 1559
QY 1560 AKINLFPALFTGCIYKALALRHYTFNNSNIDPFVYVILSIYGVLSIDIIQKX-PSPT 1619
DB 1560 AKINLFPALFTGCIYKALALRHYTFNNSNIDPFVYVILSIYGVLSIDIIQKX-PSPT 1619
QY 1620 LFRVIRLARIRIRILIRGAGIRITLLPALMMSLPALFNIGLLFLVWFYISIGMANFA 1679
DB 1620 LFRVIRLARIRIRILIRGAGIRITLLPALMMSLPALFNIGLLFLVWFYISIGMANFA 1679
QY 1680 YVKWAGIDDMFNQTFANSMCLFOITTSAGMGLSPILNTPPCDPLPLPSNSGRG 1739
DB 1680 YVKWAGIDDMFNQTFANSMCLFOITTSAGMGLSPILNTPPCDPLPLPSNSGRG 1739
QY 1740 DCGSPAVGILFFTYIIISFLIVNMVYATILLENFSVATEESTEPLESDPDMYEIWEK 1799
DB 1740 DCGSPAVGILFFTYIIISFLIVNMVYATILLENFSVATEESTEPLESDPDMYEIWEK 1799
QY 1800 FDPPEATQFIEYSVLSDFADALSEPLRIAKPNQISLIMNDLPMVSGDRITHCMDILFAFTKR 1859
DB 1800 FDPPEATQFIEYSVLSDFADALSEPLRIAKPNQISLIMNDLPMVSGDRITHCMDILFAFTKR 1859

Db 1800 FDEATQPLEYSLSDPADLSEPLIAKENQISLIMDLPMVSGDRHICMDILFAFTKR 1859
QY 1860 VGESEEMALKTOMEKEKMAAPSKISYEPIITTLARKHEEVSANVIOQAPRRHLQSS 1919
Db 1860 VGESEEMALKTOMEKEKMAAPSKISYEPIITTLARKHEEVSANVIOQAPRRHLQSS 1919
QY 1920 LKHAFLFRQOAGSGISEEDAPEREGLIAYVMSSENRPPLGPPSSSISSTSPSPYDSV 1979
Db 1920 LKHAFLFRQOAGSGISEEDAPEREGLIAYVMSSENRPPLGPPSSSISSTSPSPYDSV 1979
QY 1980 TRATSNDLVGRSGDYSHSEDLADPPSPDRRESIV 2015
Db 1980 TRATSNDLVGRSGDYSHSEDLADPPSPDRRESIV 2015
RESULT 21
ID AAR67913 standard; protein; 2019 AA.
XX AAR67913;
AC AAR67913;
XX 25-MAR-2003 (revised)
DT 05-AUG-1995 (first entry)
XX Cardiac sodium channel protein.
DE Sodium channel protein; therapeutic; diagnostic; prognostic;
KM anclarythmic; cardiant; cardioglycoside.
XX Rattus rattus.
OS Rattus rattus.
XX US5380836-A.
PN 10-JAN-1995.
XX 30-SEP-1991; 91US-00768107.
PF 13-FEB-1989; 89US-00331330.
PR (ARCH-) ARCH DEV CORP.
PA Rogart RB;
XX MPI: 1995-060381/08.
DR P-PSDB; AA081328.
XX Purified DNA's encoding rat and human cardiac sodium channel protein -
PT useful for recombinant expression to produce sodium channel proteins.
XX Disclosure; Fig 2; 39pp; English.
XX The rat cardiac channel protein has various therapeutic, diagnostic and
CC prognostic uses. It may also be used to develop more effective
CC anclarythmic, cardiant and cardioglycoside drugs. In Figure 2, the
CC sequence is compared to the deduced amino acid sequence of rat brain II
CC cDNA. (Updated on 25-MAR-2003 to correct PF field.)
XX Sequence 2019 AA;
SQ
Query Match 93.1%; Score 9767; DB 2; Length 2019;
Best Local Similarity 93.3%; Pred. No. 0;
Matches 1884; Conservative 47; Mismatches 84; Indels 4; Gaps 3;

Db 121 VRRAAVKILVHSLFMNLIWCTILTNCFVMAQHPPEWTKYVEYTFATYTFESLVKILAR 180
QY 180 GCLIAFTPLRPPMMLDSSVIMATTFEPDUNGVASARTPRVIALAKTISVIGLKT 239
Db 181 GCLIAFTPLRPPMMLDSSVIMATTFEPDUNGVASARTPRVIALAKTISVIGLKT 240
QY 240 VQALIOSVKKLADVWVLTFCGVFALIGLOFMGNLRKCYRNFALNGTNGSVADGL 299
Db 241 VQALIOSVKKLADVWVLTFCGVFALIGLOFMGNLRKCYRNFALNGTNGSVADGL 300
QY 300 VNESIDLVLSDPENYLLKNGTSDVLLCGNSDAGTCPEGYRCLKAGENDHGYTSPDPA 359
Db 301 VNSLDVLYLNDPANVLLKNGTSDVLLCGNSDAGTCPEGYRCLKAGENDHGYTSPDPA 360
QY 360 VAFALFLRLMTODCWERLYOQTLRSAGKTYMIFMLVTLGSGPYLVNLLAVYMAVEEQ 419
Db 361 VAFALFLRLMTODCWERLYOQTLRSAGKTYMIFMLVTLGSGPYLVNLLAVYMAVEEQ 420
QY 420 NOATTAEFEKKEKRFQEAEMMLKKEHEALITIRGVTVSRSSLEMSPLAVNSHERSKR 479
Db 421 NOATTAEFEKKEKRFQEAEMMLKKEHEALITIRGVTVSRSSLEMSPLAVNSHERSKR 480
QY 480 KEMSSGTECEGDRLPKSDSEDPGRAMNHLSTRLGSRYSMKRPSRSGSIPTFRRRDLGS 539
Db 481 KRLSSGTEDGDDRLPKSDSEDPGRAMNHLSTRLGSRYSMKRPSRSGSIPTFRRRDLGS 540
QY 540 EADPPADDENSTIGSEBSHRTSLVWPPLRRTSAGQPSFGTSPGHALHGKNSYTDGNG 599
Db 541 EADPPADDENSTIGSEBSHRTSLVWPPLRRTSAGQPSFGTSPGHALHGKNSYTDGNG 600
QY 600 VVSLIAGDPBATSGSHLLRPVMLBHPDPTTPEBEPGPMQLTSPACVUGFEBPGR 659
Db 601 VVSLIAGDPBATSGSHLLRPVMLBHPDPTTPEBEPGPMQLTSPACVUGFEBPGR 660
QY 660 QRALSAVSLTSALTELESRRHKPCPCWNRRLAQRVLIWECPLMNSIKGVLLVMDPPT 719
Db 661 QRALSAVSLTSALTELESRRHKPCPCWNRRLAQRVLIWECPLMNSIKGVLLVMDPPT 720
QY 720 DLTITMCIVLNTLFPALBHYNMTSEPEMLQVGNLVFTGIFPAENTFKIALDPYVYFQ 779
Db 721 DLTITMCIVLNTLFPALBHYNMTSEPEMLQVGNLVFTGIFPAENTFKIALDPYVYFQ 780
QY 780 GWNIDFSIIIVLSLMEIGLSRMSNLVSRSPFLRVFLAKSWPLNTLTIKIGNSVGL 839
Db 781 GWNIDFSIIIVLSLMEIGLSRMSNLVSRSPFLRVFLAKSWPLNTLTIKIGNSVGL 840
QY 840 GNLTLVLAIIYFIFAVVGMQLFGKNYSRLRD--SDSGLLPRWMDFFHAFILIRILCG 897
Db 841 GNLTLVLAIIYFIFAVVGMQLFGKNYSRLRISDSGLPRWMDFFHAFILIRILCG 900
QY 898 EWIETMDOMEVSGSLCLVFLVLMVIGNLVLMFLALLISFSADNLTADEDEKN 957
Db 901 EWIETMDOMEVSGSLCLVFLVLMVIGNLVLMFLALLISFSADNLTADEDEKN 960
QY 958 NIQALALRIORGIRVVKRTTMDPCGGLRORPORAALAAQOLPSCATPSPPEPTE 1017
Db 961 NIQALALRIORGIRVVKRTTMDPCGGLRORPORAALAAQOLPSCATPSPPEPTE 1020
QY 1018 KVPTRKREBEQPGQGTGDPPEPCVPIAVASEPTDQDEBDEENSLGTEBESSKOE 1077
Db 1021 KVPTRKREBEQPGQGTGDPPEPCVPIAVASEPTDQDEBDEENSLGTEBESSKOE 1080
QY 1078 SQVSGGHEPQOEPRAMQVSEETSSBAGASTSQDMWQOEKTEPOAGCCEPDPDSYE 1140
Db 1081 SQVSGGHEPQOEPRAMQVSEETSSBAGASTSQDMWQOEKTEPOAGCCEPDPDSYE 1140
QY 1138 GSTADMTATLLEQIPDQGVNDPENCFTGCVRRPCCANVTQOAPGVMMRLRKC 1197
Db 1141 GSTADMTATLLEQIPDQGVNDPENCFTGCVRRPCCANVTQOAPGVMMRLRKC 1200
QY 1198 YHIVHSWFETFIIFMILSSGALAFEDIVYEEKRTICVLEIYADKMTYVFLVEMLLK 1257
Db 1201 YHIVHSWFETFIIFMILSSGALAFEDIVYEEKRTICVLEIYADKMTYVFLVEMLLK 1260

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QY 1258 VAYGFKYFTNAMCMLDPLIVDSIVSLVANTLGFAMGPTKSTRTLRALRPLRALSRE 1317
DB 1261 VAYGFKYFTNAMCMLDPLIVDSIVSLVANTLGFAMGPTKSTRTLRALRPLRALSFE 1320
QY 1318 GMRVYVNVAVGAIPIBSIMNVAVCLIFMLIFSIIMGNVPAKRGRCINOTEGDLPLANTYIV 1377
DB 1321 GMRVYVNVAVGAIPIBSIMNVAVCLIFMLIFSIIMGNVPAKRGRCINOTEGDLPLANTYIV 1380
QY 1378 NNGSCESLNTJGELVMTKXVNFEDNVAGYIALLLQVATFGMDIMYAAVDSRGYEQP 1437
DB 1381 NNGSCESFNTGELVMTKXVNFEDNVAGYIALLLQVATFGMDIMYAAVDSRGYEQP 1440
QY 1438 QMEYNLYMYIYFVFIIFGSPFTLNLFTGVIIIDNNQOKKLGODIMTEBOKKYNYAM 1497
DB 1441 QMEDLYMYIYFVFIIFGSPFTLNLFTGVIIIDNNQOKKLGODIMTEBOKKYNYAM 1500
QY 1498 KKLGSKKPKQKPIPRPLNKYOGFIPIVTKQAFDVTIMFLCLNMVTMMVETDQSPKIN 1557
DB 1501 KKLGSKKPKQKPIPRPLNKYOGFIPIVTKQAFDVTIMFLCLNMVTMMVETDQSPKIN 1560
QY 1558 ILAKINLFFVAIFTEGCIIVKLAALRHYFTNSWNIFDFVVVILSIGTVLSDIIOKYEFS 1617
DB 1561 ILAKINLFFVAIFTEGCIIVKLAALRHYFTNSWNIFDFVVVILSIGTVLSDIIOKYEFS 1620
QY 1618 PTLFVIVILARIGRLILIRKAGKIRTLIFALMGLPALFNLGILLFVMTFYSIFGMAN 1677
DB 1621 PTLFVIVILARIGRLILIRKAGKIRTLIFALMGLPALFNLGILLFVMTFYSIFGMAN 1680
QY 1678 FAYVMEAGIDDMFNFOTFANSMLCLFOITTSAGNDGLSLPLNTGPPYCDPTLPNSNGS 1737
DB 1681 FAYVMEAGIDDMFNFOTFANSMLCLFOITTSAGNDGLSLPLNTGPPYCDPTLPNSNGS 1740
QY 1738 RGDGSSPAVGLIFFTYIIISPLIVNMVIALIILNFSVAITEESTEPLEDDFDMFYEW 1797
DB 1741 RGNCGSPAVGLIFFTYIIISPLIVNMVIALIILNFSVAITEESTEPLEDDFDMFYEW 1800
QY 1798 EKFDPEAOTFIYSVLSDPADALSEPLRIAKNQISLIMDLPMVSGGRICHMDLFAFT 1857
DB 1801 EKFDPEAOTFIYSVLSDPADALSEPLRIAKNQISLIMDLPMVSGGRICHMDLFAFT 1860
QY 1858 KRVLESSEGMALAKIOMEKEKMAANPSKISYEPIITTLRRKHEEVSAMVIOGAPRRHLLQ 1917
DB 1861 KRVLESSEGMALAKIOMEKEKMAANPSKISYEPIITTLRRKHEEVSAMVIOGAPRRHLLQ 1920
QY 1918 RSLKIASFLFRQQA-GSGISEDAPEREGLIAYVNSEFSRPLGPPSSSSISSTSPPSY 1976
DB 1921 RSLKIASFLFRQQA-GSGISEDAPEREGLIAYVNSEFSRPLGPPSSSSISSTSPPSY 1980
QY 1977 DSVTRATSDNLTQVRSQDYSHSFDLADFPSPDRDESIY 2015
DB 1981 DSVTRATSDNLTQVRSQDYSHSFDLADFPSPDRDESIY 2019
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RESULT 22

ID AAR06584 standard; protein; 2020 AA.

AAR06584;

DT 10-JAN-1991 (first entry)

DE Cardiac sodium channel.

KW Rat; arrhythmia.

OS Rattus rattus.

PN WO9009391-A.

PD 23-AUG-1990.

PF 13-FEB-1989; 89US-00310330.

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XX 13-FEB-1989; 89US-00310330.
PR (ARCH-) ARCH DEV CORP.
XX Rogart RB;
XX WPI; 1990-275095/36.
DR N-PSDB; AA005831.
XX New rate cardiac sodium channel proteins - and associated DNA sequences,
PT polypeptide(s) and peptide(s) associated with proteins, useful as
PT antiarrhythmic and cardiotoxic drugs.
XX Disclosure; Fig 2; 65pp; English.
XX The sequence deduced from cDNA derived from 3 overlapping clones, pRH-
CC 1, pRH-23, and pRH-4-31. (Deposited as ATCC 67885, 67886, and 67887
CC resp.) The clones were isolated from a cDNA library in the lambda Zap
CC vector prep. from mRNA obtd. from newborn rat hearts using rat brain II
CC cDNA probe. The protein has diagnostic, therapeutic, and prognostic
CC applications
XX
SQ Sequence 2020 AA;
Query Match 92.7%; Score 9719; DB 2; Length 2020;
Best local similarity 93.1%; Pred. No. 0;
Matches 1879; Conservative 44; Mismatches 92; Indels 4; Gaps 3;
1 MANFLPRGTSSPFRFRESLIAIEKMAEKQAR-GSTYLOESREGLPEEAPRPOLDQ 59
1 MANLLPRGTSSPFRFRESLIAIEKMAEKQARGSATSQESREGLOEEAEVRPOLDQ 60
60 ASKGLPDLXGNPPELLIGEBLEDLDPYSTOKTFTVINKKTI PRSATNALVLSPEHP 119
61 ASKGLPDLXGNPPELLIGEBLEDLDPYSTOKTFTVINKKTI PRSATNALVLSPEHP 120
120 JRRRAVILVHSLFNMILMCTILNCFVMAQHPPTKTVETFTAIYTFESIVKLAR 179
121 VRRRAVILVHSLFNMILMCTILNCFVMAQHPPTKTVETFTAIYTFESIVKLAR 180
180 GFCLHAFPLRDPNMWLDPSVLIIMAYTTEFVDLGNVALRTFRVLRALKTISVIGLKI 239
181 GFCLHAFPLRDPNMWLDPSVLIIMAYTTEFVDLGNVALRTFRVLRALKTISVIGLKI 240
240 VGALIQSVKGLADVMUTVPCLSVFLIGLQLEPMGNLRHCVRNFTLNGTNSVEADGL 299
241 VGALIQSVKGLADVMUTVPCLSVFLIGLQLEPMGNLRHCVRNFTLNGTNSVEADGL 300
300 VMSLIDYLDSPENYLLKNGTSDVILCGNSSDAGTCPEGYRCLAKGENPDHGYTSPDFA 359
301 VMSLIDYLDSPENYLLKNGTSDVILCGNSSDAGTCPEGYRCLAKGENPDHGYTSPDFA 360
360 WAFPLAFRLMTQDCWERYOQTLSAGKIYIFPMVLVIFLGSFYLVNLLIIVAVMAYEQ 419
361 WAFPLAFRLMTQDCWERYOQTLSAGKIYIFPMVLVIFLGSFYLVNLLIIVAVMAYEQ 420
420 NOATTAETEEKERKFOEMEMTLKKEHELTTRGVDTYSRSLSEMSPLAPVNSHERRSKR 479
421 NOATTAETEEKERKFOEMEMTLKKEHELTTRGVDTYSRSLSEMSPLAPVNSHERRSKR 480
480 KRMSSGTEECGEDRLPKSDSEDDGRANMHLISLRTGLSRTSKPKPSRGSITFFRRDLGS 539
481 KRISSTEDGDDRLPKSDSEDDGRANMHLISLRTGLSRTSKPKPSRGSITFFRRDLGS 540
540 EADPADDENSTAGESSESHRTSLVWPPLRRTSAQGPSPGTSAFGHALHKKNSVTDGNG 599
541 EALVPWPLRHSQAQESHRTSLVWPPLRHSQAQGPSPGTSAFGHALHKKNSVTDGNG 600
600 VVSLIAGDPPAATPGSHILARPVNLHPPDPTTSSERPGGQMLTQAPCDVGEFPGAR 659
601 VVSLIAGDPAATPGSHILARPVNLHPPDPTTSSERPGGQMLTQAPCDVGEFPGAR 660
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QY 660 QRLASAVSLTSALEELBSRHKCP CWNRLAORYLWECCPLMNSIKQVVLVYMDPT 719
Db 661 QRLASAVSLTSALEELBSRHKCP CWNRFQOHLIWECCPLMNSIKQVYFVMDPFA 720
QY 720 DLTIMCVLNTLFNALHYNMTSFEEMLOVGNLVFTGIFPAENTFKI IADPYTYRQO 779
Db 721 DLTIMCVLNTLFNALHYNMTSFEEMLOVGNLVFTGIFPAENTFKI IADPYTYRQO 780
QY 780 GNNPDSIIIVLSLMBELGSRMSNLSVRSFRLVFPFLAKSWPNTLTIKIGSVGL 839
Db 781 GNNPDSIIIVLSLMBELGSRMSNLSVRSFRLVFPFLAKSWPNTLTIKIGSVGL 840
QY 840 GNLTLVLAIVFI FAVVGWQLFGKNYSBLR--SDSGLLPRXMMDFHAFILIRILCG 897
Db 841 GNLTLVLAIVFI FAVVGWQLFGKNYSBLRHSISGGLPRXMMDFHAFILIRILCG 900
QY 898 EMIETMDOMEVSGSLCLVFLVMTIGNLVNLPLALLISFSADNLTAPDEDRNN 957
Db 901 EMIETMDOMEVSGSLCLVFLVMTIGNLVNLPLALLISFSADNLTAPDEDRNN 960
QY 958 NLQIALARIORGLRFRKRTWDFCCGLRORPQRAALAAOGLRSCATPYSPPPETE 1017
Db 961 NLQIALARIORGLRFRKRTWDFCCGLRORPQRAALAAOGLRSCATPYSPPPETE 1020
QY 1018 KVPTRKRETRFEGBQPGCGTGPDEPVCPIA VASDPTDOEDDEENSLGTEESSKOE 1077
Db 1021 KVPTRKRETRFEGBQPGCGTGPDEPVCPIA VASDPTDOEDDEENSLGTEESSKOE 1080
QY 1078 SQPVSGGPEAPDSHTSQVSATASSEBASASQDMRQOWKAEPOARCGGTPEBSCSE 1137
Db 1081 SQPVSGGHEPQOEPRAMQVSETTSSEAGASTSQADMQOEGTBEPOARCGGTPEBSCSE 1140
QY 1138 GSTADMTTALHEBQIPDLGQDVKDPEDCFTEGCRRCPCCAVDTQOAGKVMWLRKTC 1197
Db 1141 GSTADMTTALHEBQIPDLGQDVKDPEDCFTEGCRRCPCCAVDTQOAGKVMWLRKTC 1200
QY 1198 YHIVHSWFEFTIIMILSSGALAFEDIYLEERKTIKYLEYADKMTYVYVLEMLKM 1257
Db 1201 YHIVHSWFEFTIIMILSSGALAFEDIYLEERKTIKYLEYADKMTYVYVLEMLKM 1260
QY 1258 VAYGFKKYFTNAMCWLDFLIYDVSLVSLVANTLGAEMGPIKSLTURLALRPLRSLRE 1317
Db 1261 VAYGFKKYFTNAMCWLDFLIYDVSLVSLVANTLGAEMGPIKSLTURLALRPLRSLRE 1320
QY 1318 GMRVYVNALVGAIPBIMNVLVCLIFPLIFSIIMGVNLPAKRGRCINOTEGDPLANTYIV 1377
Db 1321 GMRVYVNALVGAIPBIMNVLVCLIFPLIFSIIMGVNLPAKRGRCINOTEGDPLANTYIV 1380
QY 1378 NNSKQCESLNTLGEIYMTKVKVNPDPNGAGYALALQVATPFKGMNDIMYAAVDSRGYEROP 1437
Db 1381 NNSKQCESLNTLGEIYMTKVKVNPDPNGAGYALALQVATPFKGMNDIMYAAVDSRGYEROP 1440
QY 1438 QMEVNLVYVFI FFIIFGSEFTLNLFTGVIIIDNFOOKKGLGQGDIMTEBOKKYNYAM 1497
Db 1441 QMEVNLVYVFI FFIIFGSEFTLNLFTGVIIIDNFOOKKGLGQGDIMTEBOKKYNYAM 1500
QY 1498 KKLGSKKPQKPIPRPLANKYQGFIDIVTKQAPDVTIMFLICLNNYTMVETDOSPCKIN 1557
Db 1501 KKLGSKKPQKPIPRPLANKYQGFIDIVTKQAPDVTIMFLICLNNYTMVETDOSPCKIN 1560
QY 1558 ILAKINLFFVAIFPGEICIVKLAAALRHYFTNSWNI FDDVVYVLSIGVTLSDIIOKXFFS 1617
Db 1561 ILAKINLFFVAIFPGEICIVKLAAALRHYFTNSWNI FDDVVYVLSIGVTLSDIIOKXFFS 1620
QY 1618 PTLFRVIRIARIGRLIRLIRGAKGIRTLIFALMMSLPALFNIGLILFLMFIYSIFGMAN 1677
Db 1621 PTLFRVIRIARIGRLIRLIRGAKGIRTLIFALMMSLPALFNIGLILFLMFIYSIFGMAN 1680
QY 1678 FAYYKMEGIDMFEFQTFANSMCLFOITTSAGNDGLSLPLANTGPYCPPLTPNSNGS 1737
Db 1681 FAYYKMEGIDMFEFQTFANSMCLFOITTSAGNDGLSLPLANTGPYCPPLTPNSNGS 1740
QY 1738 RGDGSPAVGLIFFTYIIISFLVVMNYIAIILENFVATBESTEPLSEDDPDMFYETW 1797

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Db 1741 RNCGSPAVGLIFFTYIIISFLVVMNYIAIILENFVATBESTEPLSEDDPDMFYETW 1800
QY 1798 EKPDPEAQOFIEYSVLSFPALASBPRLAIAPNOSLNNMLPMVSGRIRHMDILFAPT 1857
Db 1801 EKPDPEAQOFIEYSVLSFPALASBPRLAIAPNOSLNNMLPMVSGRIRHMDILFAPT 1860
QY 1858 KRIVGESGEMDALKIOMEKEMANPSKISYEPIITTLRRKHGEVSAMVIORAFRRHLQ 1917
Db 1861 KRIVGESGEMDALKIOMEKEMANPSKISYEPIITTLRRKHGEVSAMVIORAFRRHLQ 1920
QY 1918 RSLKRAEFLFRQOA--GSGLSEEDAPEREGLIAYVWSENFSPPLGPPSSSSISSTSPPSY 1976
Db 1921 RSVKRAEFLFRQOAGSGSLSEEDAPEREGLIAYVWSENFSPPLGPPSSSSISSTSPPSY 1980
QY 1977 DSVTRATSDNIOVRGSDYSHSEDLADPPSPDRDRESIV 2015
Db 1981 DSVTRATSDNIOVRGSDYSHSEDLADPPSPDRDRESIV 2019

RESULT 23
AAU19518
ID AAU19518 standard; protein, 1603 AA.
XX
AC AAU19518;
XX
DT 04-DEC-2001 (first entry)
XX
DE Human diagnostic and therapeutic polypeptide (DITP) #104.
XX
KW Human; receptor; diagnostic; therapeutic; gene therapy; vaccine;
KW cell proliferator; disorder; Crohn's disease; lymphoma; leukaemia;
KW acquired immune deficiency syndrome; AIDS; autoimmune disorder;
KW respiratory disorder.
XX
OS Homo sapiens.
XX
PN WO200162927-A2.
PD 30-AUG-2001.
XX
PF 21-FEB-2001; 2001WO-US006059.
XX
PR 24-FEB-2000; 2000US-0184631P.
PR 24-FEB-2000; 2000US-0184637P.
PR 24-FEB-2000; 2000US-0184638P.
PR 24-FEB-2000; 2000US-0184768P.
PR 24-FEB-2000; 2000US-0184769P.
PR 24-FEB-2000; 2000US-0184770P.
PR 24-FEB-2000; 2000US-0184771P.
PR 24-FEB-2000; 2000US-0184772P.
PR 24-FEB-2000; 2000US-0184773P.
PR 24-FEB-2000; 2000US-0184774P.
PR 24-FEB-2000; 2000US-0184776P.
PR 24-FEB-2000; 2000US-0184777P.
PR 24-FEB-2000; 2000US-0184778P.
PR 24-FEB-2000; 2000US-0184797P.
PR 24-FEB-2000; 2000US-0184813P.
PR 24-FEB-2000; 2000US-0184837P.
PR 24-FEB-2000; 2000US-0184841P.
PR 24-FEB-2000; 2000US-0185213P.
PR 24-FEB-2000; 2000US-0185216P.
PR 24-FEB-2000; 2000US-0185219P.
PR 12-MAY-2000; 2000US-0203785P.
PR 15-MAY-2000; 2000US-0204226P.
PR 16-MAY-2000; 2000US-0204525P.
PR 16-MAY-2000; 2000US-0204821P.
PR 16-MAY-2000; 2000US-0204908P.
PR 16-MAY-2000; 2000US-0205232P.
PR 17-MAY-2000; 2000US-0204815P.
PR 17-MAY-2000; 2000US-0204863P.
PR 17-MAY-2000; 2000US-0205221P.
PR 17-MAY-2000; 2000US-0205285P.
PR 17-MAY-2000; 2000US-0205286P.
PR 17-MAY-2000; 2000US-0205287P.

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DB 1431 IVNNKSDCKIONSTGSPFWNVNKNFNDVAMGYLALLQVATPKGMMWIMYAVDSREYVM 1490

QY 1436 OPQWENLYMYTYFYFIIFGSPFTLNLPIGVIIIDNPNQKKGLGGODIFMTEBOKKYN 1495

DB 1491 QPKMDNVMYMYFYFIIFGSPFTLNLPIGVIIIDNPNQKKGLGGODIFMTEBOKKYN 1550

QY 1496 AMKKLGSKKKPKQKPIPRPLNKYGFIEDIVTKQAFDVITIMFLICLMMV 1542

DB 1551 AMKKLGSKKKPKQKPIPRPLNKYGFIEDIVTKQAFDVITIMFLICLMMV 1597

RESULT 24

ID ADP79541 standard, protein; 2000 AA.

AC ADP79541;

XX 04-NOV-2004 (first entry)

DT 04-NOV-2004 (first entry)

DE Human sodium III channel splice variant (hna1118).

XX Human, sodium III channel; hna1118; splice variant; antiarrhythmic;

KM analgesic.

OS Homo sapiens.

XX MO2004050857-A2.

PN 17-JUN-2004.

PD 04-DEC-2003; 2003WO-US038796.

XX 04-DEC-2002; 2002US-0431794P.

PR (EURO-) EUROCELLTQUE SA.

XX Kammesheidt A, Hodges D;

PI MPI: 2004-450725/42.

DR N-F8DB; ADP79540.

XX New human sodium III channel, useful in treating cardiac arrhythmias,

PT herpes virus infection, diabetes mellitus, or vasculitis.

PS Claim 9; SEQ ID NO 2; 101pp; English.

XX The present sequence is that of a novel splice variant of the human

CC sodium III channel alpha subunit, denoted hna1118. The cDNA was cloned

CC by RT-PCR from human embryonic brain total RNA using human Na111 specific

CC primers ADP79546-ADP79547. The hna1118 sequence contains an additional

CC 49 amino acid residues that do not appear in a previously reported human

CC Na111 ADP79543, and also differs from a previously reported splice

CC variant of human sodium channel alpha subunit ADP79545 by 12 amino acids

CC out of 2000. Transient transfection of HEK293 cells by hna1118 resulted

CC in expression of functional sodium channels. The invention provides:

CC hna1118 proteins and their fragments and derivatives; hna1118-encoding

CC nucleic acids and their fragments, including primers, probes, and

CC regulatory sequences; hna1118-specific antibodies; and methods of using

CC these materials to detect the presence of hna1118 proteins or nucleic

CC acids. The invention also provides an assay method for screening to

CC identify selective modulators of hna1118 expression or activity. These

CC may be useful for treatment of conditions associated with sodium channel

CC over- or under-expression, e.g. for treatment of cardiac arrhythmias,

CC neuronal disorders, nociceptive pain-related diseases and neuropathic

CC pain-related diseases, e.g. pain from peripheral nerve trauma, herpes

CC virus infection, diabetes mellitus, causalgia, plexus avulsion, neuroma,

CC limb amputation or vasculitis.

XX Sequence 2000 AA;

QY 5 LPRGTSFRRFTRESLAIEKMAEKOARGSTTLQESREGIPREEARPOLDLQASKL 64

DB 6 LVPGPBESFRFTRESLAIEKMAEKAKEAKKPKKEQDN-----DDEKKKXPNSDLEAGNKL 61

QY 65 PDLYGNPPOELIGELEDLDPPEYSTOKTPIVANKGTIFRPSANALVYLSFPHIRRAA 124

DB 62 PPIYGDIDPEWVSEPLEDLDEYYINKKTFIVNKKGAIFRPSATSALYILFPLNIVRKIA 121

QY 125 VKILVHSLFNNLINCTILTNCFVMAQHDPPEWTKVETTFPAIYFESLVITLAFGLCH 184

DB 122 IKILVHSLFNNLINCTILTNCFVMAQHDPPEWTKVETTFPAIYFESLVITLAFGLCH 181

QY 185 APTFLRDPNNLDPFVITMAVTEFVDIGNVSALRTFVPLAKTISYISGLKTVGALI 244

DB 182 DFTFLRDPNNLDPFVITMAVTEFVDIGNVSALRTFVPLAKTISYISGLKTVGALI 241

QY 245⁴QSVKGLADVWVLYVFCLSVFPALIGLQFMGNLRHKVCR-----NFTAL----- 287

DB 242 QSVKGLADVWVLYVFCLSVFPALIGLQFMGNLRHKVCR-----NFTAL----- 287

QY 288 NGTNGSVADGLWESLDLYSDPENYLLKNGTSDVLLCGNSSPDGTCPEGRCIKAGEN 347

DB 302 NGTFVNVYTMSTFNKMD---YIGDSSHFYVLDGQDPLLCGNGSDAGQCPBGYICVKAQRN 358

QY 348 PDHGYTSPDSFAMAPALFRLMTODCWERLYQOQLRSAGKLYMIFPMLVIFAGSPYVNL 407

DB 359 PNYGYTSPDYSMAFLSLFRLMTODYENLQULIRAKGKTYMIFVLYVIFLGSFYVNL 418

QY 408 ILAVVAMAVERONQATTAETEKEKRFQEMEMLKKEHALT-----IRGV 453

DB 419 ILAVVAMAVERONQATTAETEKEKRFQEMEMLKKEHALT-----IRGV 453

QY 454 DTVSRSSLEMSPLAPVNSHERSKRRKMS-----SGTECEGDLPKXSDSDGPPAMHL 509

DB 479 GELLSSSESEAKLSKSAKEWRNRKRRRREHLEGNKKGSRDSPKSESSESVRSFL 538

QY 510 SLTRGLSRTSMKP-----RSRGSIFTFRRR--DLGEADPADEN 548

DB 539 FSDGNRLTSDKCKCSPHQSLSTIRGSLFSRRRSKTSIFSGRGAQVGSBNDPADEN 598

QY 549 STAGESESHRTSLVP--WPLRRTSAQGPSPGT--SAPGHALGKNSVDNCGVSLIG 605

DB 599 STFDSESRRLDPLVPHRGERRNSVNSQAMSRMVGLPANGMHSITVDCNGVSLIG 658

QY 606 AGDPBAPSPGSHLRPVMLEHPPTTPPSBEGPQMLTSAQPCVDGEEFGARALSA 665

DB 659 -GSPALTSPTQL-----PPEGTT--TEVEKRRRLSSYQISMEMLEDSGRRAVSI 708

QY 666 VSVLTSLMEERESRHCPCPCWNLQAOXYLIWECCPLMMSIKQVYKLVMDPFTDLTITM 725

DB 709 ASILTNTVEERESRCKPCPCWNLQAOXYLIWECCPLMMSIKQVYKLVMDPFTDLTITM 768

QY 726 CIVANTLPMALHNMTSEFEEMLOVGNLVTGTIFTAMTKIALLDPYVYFOQGMNIFD 785

DB 769⁴CIVANTLPMALHNMTSEFEEMLOVGNLVTGTIFTAMTKIALLDPYVYFOQGMNIFD 828

QY 786 SIIVLSLMEIGLSRMSNLVLRSPFLIRVFKLAKSWPTLNTLKIIGNSVGAIGNLTIV 845

DB 829 GIIVLSLMEIGLSRMSNLVLRSPFLIRVFKLAKSWPTLNTLKIIGNSVGAIGNLTIV 888

QY 846 LAIVVFAVVGMLPGKNYSE--LRDSDSLPRMNMDFPHAFLIFRILICEWERTM 903

DB 889 LAIVVFAVVGMLPGKNYSE--LRDSDSLPRMNMDFPHAFLIFRILICEWERTM 948

QY 904 WDCEVSGOSLCLVFLVAVTGNLVVNLFTALLLSFSADNTLAPDEBEMNLTQAL 963

DB 949 WDCEVAGQTMCLVFLVAVTGNLVVNLFTALLLSFSADNTLAPDEBEMNLTQAL 1008

QY 964 ARIGRLRFVKKRTTWDFCCGLLRORPQKPAALAAQGLPSCIATPYSPPEPEKVPETR 1023

DB 1009 GRMKGIDIVYNNKRE--CFQKAPFRKPKVIEIHGNKIDSCMSNNTG-----TEISKEL 1061

```
QY 1024 KETPEBEGQPGQCPGDP-----PVCPIAVAESPTDDQEDSEN 1065
Db 1062 NYLIDGNGTTSVGSGSSVEKXYIDENDYMSFINNPISLTVPVPLAVGSDPE----- 1113
QY 1066 SLGTEEESKQESQPSGSGPEAPPDRTWSQVSATASABASASQADMRQWKAEPQAP 1125
Db 1114 NLNTEEFSESELE-----EKEKLNANS----- 1138
QY 1126 GCGETPEDSCSEGSTAD--MTNTAELHQIDPLGDVDPEDCFTEGCVRCPCCAVDTT 1183
Db 1139 -----SEGSTVDVLPREGQAEETPE--EDLK-PEACFPEGCIKKFPQCVSTE 1185
QY 1184 QAPGVWRLKRTCTCHIEHSEHFEPIIFMLISGALAFEDYIEBKRTIKVLEEVADK 1243
Db 1186 EGKGIMWNLRKTCYSIEHWMFETIVFMILSSGALAFEDYIEBKRTIKVLEEVADK 1245
QY 1244 METYVFLVEMLLKWAAYGFKKFTNAMCMLDFLIDVSLVSLVANTLGLFAEWGPIKSLRT 1303
Db 1246 VFTYIFILEMLLKWAAYGFGQYFTTAKCMLDFLIDVSLVSLVANTLGLFAEWGPIKSLRT 1305
QY 1304 LRALRPLRALSFEGRVAVVAVLGAIPSIMNVLVCLIFMLISIMGNVLPAGKFGRCI 1363
Db 1306 LRALRPLRALSFEGRVAVVAVLGAIPSIMNVLVCLIFMLISIMGNVLPAGKFGYHCV 1365
QY 1364 NOTBEDLPLNTIYVNNKSCQESLNTGELVYTKVYVNPVNGAGYALLQVATPEGMDI 1423
Db 1366 NMTGTNM--FDISDVNNLSDCQALG--KQARMKNVAVNPVNGAGYALLQVATPEGMDI 1422
QY 1424 MYAAVDSRGVEPOPOMEVNLVWYIFVFIIFGSEFTLNLFGVLIIDNFOQKKLGGSD 1483
Db 1423 MYAAVDSRDVQLQPYEENLVWYLFVFIIFGSEFTLNLFGVLIIDNFOQKKLGGSD 1482
QY 1484 IFMTBEOKKYVNAKKLGSKKPKPIPRPLNKYQCFIPDIYTKQAFDVTYIFLILANVT 1543
Db 1483 IFMTBEOKKYVNAKKLGSKKPKPIPRPLNKYQCFIPDIYTKQAFDVTYIFLILANVT 1542
QY 1544 MMVETDDQSPKINILAKINILFVAFGECECVKLAALRHYFTSMNIPDVVVILSIV 1603
Db 1543 MMVETDDQSKWTLVLSRINLVFVAFGECECVKLAALRHYFTSMNIPDVVVILSIV 1602
QY 1604 GTVLSDIIOKYPFSPFLFVRILARIGRILIRAKAGIRTLFALMMSLPALFNIGILL 1663
Db 1603 GMFLAMEIEKTVSPTLRVITLARIGRILIRAKAGIRTLFALMMSLPALFNIGILL 1662
QY 1664 FLVMEFYISFGKAPVAVYKMEAGIDMENEQTFPANSMLCFQITTSAGDGLSLPLTNG 1723
Db 1663 FLVMEFYIAIFGMSNPAVYKMEAGIDMENEQTFPANSMLCFQITTSAGDGLSLPLTNSA 1722
QY 1724 PRCYCP--TLPNNGSRGDCGSPAVGILFETTYIIISFLVVMYTAIIENFSVATEEST 1782
Db 1723 PRCYCP--TLPNNGSRGDCGSPAVGILFETTYIIISFLVVMYTAIIENFSVATEEST 1782
QY 1783 EPLSDDDFMPEYIEKEPPEATQFLEYVLSDFADALSEPLIAKPNQISLINDLPMV 1842
Db 1783 EPLSDDDFMPEYIEKEPPEATQFLEYVLSDFADALSEPLIAKPNQISLINDLPMV 1842
QY 1843 SCDRIHCDILFAFTKRVLVGSGEMDALKIOMEKFMANPSKISYEPTITTLRKHEEV 1902
Db 1843 SCDRIHCDILFAFTKRVLVGSGEMDALKIOMEKFMANPSKISYEPTITTLRKHEEV 1902
QY 1903 SAMVYQARFRRLRLQSLKGLASFLPRQOQSGLSGEDABERBGLAYVWSENFSPRLGP 1962
Db 1903 SAMVYQARFRRLRLQSLKGLASFLPRQOQSGLSGEDABERBGLAYVWSENFSPRLGP 1962
QY 1963 SSSSISSTSPSPSYDSVTRATSDNLC 1988
Db 1963 SSSSISSTSPSPSYDSVTRATSDNLC 1988
QY 1987 KTDGSSSTSPSPSYDSVTRATSDNLC 1982
Db 1987 KTDGSSSTSPSPSYDSVTRATSDNLC 1982
```

```
AC AAB99676.4
XX
XX 04-SEP-2001 (first entry)
DT
XX
XX Human adult form of SCN2A protein sequence SEQ ID NO:35.
DE
XX
XX Human; epilepsy; chromosome 2; SCN1A; SCN2A; SCN3A; identification;
KW diagnosis; mutation; chromosome 2q23-q31; neurological disorder;
KM anticonvulsant; neuroprotective.
XX
XX Homo sapiens.
XX
XX MO200138564-A2.
XX
XX 31-MAY-2001.
XX
XX 24-NOV-2000; 2000WO-CA001404.
XX
XX 26-NOV-1999; 99US-0167623P.
XX
XX (UYMC-) UNIV MCGILL.
PA
XX
XX Rouleau GA, Latreuniere RG, Rochefort D, Cossette P, Rasedale D;
PI N-PDB; AAH55793.
DR
XX
XX WPI; 2001-355945/37.
XX
XX N-PDB; AAH55793.
PT
XX
XX Determining a predisposition to epilepsy and/or development of epilepsy
PT comprises determining the genotype of SCN1A, SCN2A and/or SCN3A, or a DNA
variant, equivalent, or mutation which shows a linkage disequilibrium.
PS Disclosure: Page 123-130; 268pp; English.
XX
XX The present invention describes a method (M1) of determining an
XX individual's predisposition to epilepsy and/or development of epilepsy,
XX as well as predicting the individual's response to medication. The method
XX comprises determining the genotype of at least one gene selected from
XX SCN1A, SCN2A or SCN3A, or a DNA variant, equivalent, or mutation which
XX shows a linkage disequilibrium. SCN1A, SCN2A and SCN3A are all sodium
XX channel genes located on chromosome 2. The idiopathic generalised
XX epilepsy (IGE) gene is more specifically localised on chromosome 2q23-
XX q31. Compounds identified as modulators of the biological activity of
XX SCN1A, SCN2A or SCN3A proteins or genes, are useful for treating epilepsy
XX or other neurological disorders. They have anticonvulsant and
XX neuroprotective activities. AAH55763 to AAH56164 and AAB99674 to AAB99679
XX represent SCN1A, SCN2A, and SCN3A cDNAs, gene fragments, PCR primers,
XX oligonucleotides and proteins given in the exemplification of the present
XX invention
XX
XX Sequence 2005 AA:
SQ
Query Match 61.0%; Score 6394.5; DB 4; Length 2005;
Best Local Similarity 62.2%; Pred. No. 0;
Matches 1301; Conservative 231; Mismatches 360; Indels 201; Gaps 33;
QY 5 LLPGTSFRFRFTESLAIKEMAEKQARSTTLQESREGLPBEAPRPOLDLOASKKL 64
Db 6 LPPEPDSFRFRFTESLAIKEMAEKQARSTTLQESREGLPBEAPRPOLDLOASKKL 62
QY 65 PDLYGNPQOEILGEBLEDLDPFYSTOKTFVILNKGKTIIFRSATNALVYVSPFPIRRA 124
Db 63 PFIYODIPPEVAVSPLBEDLDYIYINKKTFVILNKGKTIIFRSATNALVYVSPFPIRRA 122
QY 125 VKILVHSLFNNLIMCTIITNCVEMAQNDPPWTYKVEYTFPAITYFESLVKILARGFLH 184
Db 123 IKILVHSLFNNLIMCTIITNCVEMAQNDPPWTYKVEYTFPAITYFESLVKILARGFLH 182
QY 185 APTFLRDWNNLDPESVIIMAYTTEFPVDLGNYSALRTFPLVLAALKTISYSGKTIYVGLI 244
Db 183 DFTFLRDWNNLDPESVIIMAYTTEFPVDLGNYSALRTFPLVLAALKTISYSGKTIYVGLI 242
QY 245 QSVKTLADVMVLYVFCLSVFAVLIIGLQFMGNLRHKCV-----NFTLANGTNGSV 294
Db 245 QSVKTLADVMVLYVFCLSVFAVLIIGLQFMGNLRHKCV-----NFTLANGTNGSV 294
```

RESULT 25
AAB99676
ID AAB99676 standard; protein; 2005 AA.
XX

Dh	243	QSVKSLSDVMILTVFCLSPALIGLOTFMGNLRKNCQMPPNDSSEINIT5F--FNNSL	300
Qy	295	EADGLVWB-----SLDVLYPDENYLLKNGTSDVLLCGNSSDAGTCBEGYCLRAGNP	348
Dh	301	DONGTFFNRVTSI FNMDEXIEDKSHFYFLEQNDALLCGNSSDAGCPEGYICVAGRNP	360
Qy	349	DHGTSFSPSFAAFATLPRIMTQDCWERLYOOTLASAGLTYMIFMLVYFPLGSFVLVNL	408
Dh	361	NYGYSFDTFSAWAFSLFRLMTQDPEWENLYOULLTAAGCTYMI FPLVYFIDSFYLINLI	420
Qy	409	LAVNMAVEBQOATIAETBEKCFQEAEMMLKKEHEALTR-----GV	453
Dh	421	LAVNMAVEBQOATIEEAEQEAEPQML EOLKQOEBAQAAAAAASDPSBAGGI	480
Qy	454	DTVRSLSLEMSPLAVNLSHE--RRSKRRKMSGCTEBCEDRLPKXSDEGPR-----	504
Dh	481	GVFSESSVASXLSKSEKELKNRRKXKQKQEGEBE-KNDRVLLKSESDSIRRKGRF	539
Qy	505	-----AMNHLSTRGLSRTSKKPRSSGSI FTFRRR--DIASEADPDDENS	549
Dh	540	SLEGRLLTYEKRFSSPHOSLSTRGSLPSPRNNSASLPSFGRKADIGSEMDPADDEHS	599
Qy	550	TAGESHSRTSLV--WPLRRTSAQOGSPGTSA-PGHALGKXNSTYDCNGVYSLGA	606
Dh	600	TFEDNDSRDSLFVHRHGERHSHVSOASRSARVLPIIPMNGKMSAVDCGVVSLG-	658
Qy	607	GDPKATSPGSHLRVEMLEHPDITTPSEBPGPOMLTQAPCVDFEEPGARQALNAV	666
Dh	659	GPSTLSAQGL-----PGTTTETEL--RRKRSYHNSMLDEPSTRQAMSLA	708
Qy	667	SVLTSALBELBSRHKCPRCNNRLAQRYLIECCPLMMSIKQGVKLVMDPPTDITTWC	726
Dh	709	SILTMTBELEBSRCKCPWCWKFMNMCIMCCPMVKVXLVNLVMDPVDALATITC	768
Qy	727	IVLNTLPMALBHYNNTSEEBMLQVGNVFTGIFLAEMTEFKIALDPYYYPQGNINDS	786
Dh	769	IVLNTLPMABEYPTMTEQSSVLSGVLNFTGIFLAEMFLKIADPYTTFQEGNNITDG	828
Qy	787	IIVLSLMEIGLSRMSNLSVLSRFFLLRVEFKLAKSPMLNTLTIKIGNSVGLAGMLTVL	846
Dh	829	FIVSLSLMEIGLANEGLSVLSRFFLLRVFKLAKSPMLNTLTIKIGNSVGLAGMLTVL	888
Qy	847	AIIVFIFAVNGMOLFKNYS--LRDSGGLPRHNMDFPHAFILIFRILGEMIEETW	904
Dh	889	AIIVFIFAVNGMOLFKNYSKVCVCKISNDCELPRHMHDFPHFIFIVRIGEMIEETW	948
Qy	904	AIIVFIFAVNGMOLFKNYS--LRDSGGLPRHNMDFPHAFILIFRILGEMIEETW	904
Dh	949	DOHEVSGSLCLVGLVMVGNVNLFLALLSSFSADNLTPDDRENNNLQIALA	964
Qy	965	RIOGRLARVAKRTTWFCCGLLRORQOKPAL-----AAQOLPSCIATPYSPPEPTE	1011
Dh	1009	RMQKQIDPVKKIRF--IQKAPRKQKALDEIRPLEDLNKKDCSIS--	1058
Qy	1018	KVPPTRKTRPEEGE--OPGGTP-----GDPE-PVCVPIA	1056
Dh	1055	-----NHTTIEIGDNLVNLKQNGTTSIGISVEKYYVDESDYMSFINNPSTVTVPIA	1108
Qy	1051	VASPTDQOEDEBENS LGTEBESSKQESQOPVSGGEAPDPDRKMSQVATASSPAASAS	1110
Dh	1109	VGEISDFE-----NLNTEBSSSDME-----ESKCKLNATSSSEST--	1145
Qy	1111	QADWRQKAPQAPGCGEPEDSCSBGSTADMTATLLEQIPDLAQDVDPDEPCFTGEG	1170
Dh	1146	-----VIGAPABEGEP-----VEPESLE-----PEACFTED	1174
Qy	1171	CVRRCPCCAVDTTQAPGVMMRLRKYCHIVHSNFTFIIFMILLSSGALAFEDIVALE	1230
Dh	1175	CVRRKCCQGISIEBKGLMMNLRTKYCYIVHNHFEFIFVMIILLSSGALAFEDITYEQ	1233
Qy	1231	RKTIKVLLEVDKMTYFVLEMLKMWAVGPKKFTPAKACMLDPLIDVYSVLSLVANTL	1296
Dh	1235	RKTIKTMLEVDKMTYFVLEMLKMWAVGQVYFTPAKACMLDPLIDVYSVLSLVANTL	1294

QY	1291	GAEMGPRKISRTLRALRPLRALSREFGEMRVVNAALVCAISIMNVLLVCLIFMLIRSIM	1350
Db	1295	GYSLEGAIKSIRTLRALRPLRALSREFGEMRVVNAALVCAISIMNVLLVCLIFMLIRSIM	1354
QY	1351	GVNLFAGKFGKGINOTEGDLPLNTTIVNKSQCESL--NLTGELVMTKVAVNEDNVAG	1407
Db	1355	GVNLFAGKFGKGINOTEGDLPLNTTIVNKSQCESL--NLTGELVMTKVAVNEDNVAG	1411
QY	1408	YLALLQVATFKGMADIMTAAVDSRGYEBOPQWYNYLYFYVFIIRGSEFTLNLFIGV	1467
Db	1412	YLSTLQVATFKGMADIMTAAVDSRGYEBOPQWYNYLYFYVFIIRGSEFTLNLFIGV	1471
QY	1468	IIDNPNOOKKDLGGODLFMTBEOKKYYNAMKSGSKPKQKPIRPLNKYGQFIDITYKQ	1527
Db	1472	IIDNPNOOKKDLGGODLFMTBEOKKYYNAMKSGSKPKQKPIRPLNKYGQFIDITYKQ	1531
QY	1528	AFDVTYIMELCLANNVTMMVETDDOSPKEKINILAKINLFFVAIFTEGECIVKLAALRHYFT	1587
Db	1532	VFDISIMILICLANNVTMMVETDDOSPKEKINILAKINLFFVAIFTEGECIVKLAALRHYFT	1591
QY	1568	NSKNIIPDFVVVILISIVGTVLSDDITQKTFPSFTLFRVYRLARIGILRLIRGAKGIRTLF	1647
Db	1592	IGNNIPDFVVVILISIVGTVLSDDITQKTFPSFTLFRVYRLARIGILRLIRGAKGIRTLF	1651
QY	1648	ALMMSPLPLFNIGLLPLFNMFTYSIFGMANPAVYKMEAGIDDMNPOTPANSMCLPQIT	1707
Db	1652	ALMMSPLPLFNIGLLPLFNMFTYSIFGMANPAVYKMEAGIDDMNPOTPANSMCLPQIT	1711
QY	1708	TSAGMDGLSPILNTGPEPCDPTLPSNGS--RGDCGSPAVGILFTTYIITSFLIVNMY	1766
Db	1712	TSAGMDGLSPILNTGPEPCDPTLPSNGS--RGDCGSPAVGILFTTYIITSFLIVNMY	1771
QY	1767	IAILLNFSVATBESTEPLSEDDPDMEYELWEKEDPEATQPIEKVSUSDPAALSEPLRI	1826
Db	1772	IAVILNFSVATBESTEPLSEDDPDMEYELWEKEDPEATQPIEKVSUSDPAALSEPLRI	1831
QY	1827	AKPNQISLIMNDLPMVSGDRHCHMDILFAFTKRVLGRSGGMDALKIOMEKFFMANANSKI	1886
Db	1832	AKPNQISLIMNDLPMVSGDRHCHMDILFAFTKRVLGRSGGMDALKIOMEKFFMANANSKI	1891
QY	1887	SYEDITTTTLRKKEEVSAMVYIORAFRSHLLORSILKHASPLFRQOAGSGLSBEDAPEREGL	1946
Db	1892	SYEDITTTTLRKKEEVSAMVYIORAFRSHLLORSILKHASPLFRQOAGSGLSBEDAPEREGL	1949
QY	1947	IAYVMSENFSPRLCPBSSSSISSTSPSPSIDSVTRATSDMLQVRGSSYSHSED	1999
Db	1950	LIDLINENST---PEKTDMTPTSTTSPSYSDSVTKPEKEKFE--KDKSEKED	1995
RESULT 26			
ID	ADY27148	ADY27148 standard; protein; 2005 AA.	
AC	ADY27148;		
DT	05-MAY-2005	(first entry)	
DE	Human SCN2A variant R223Q.		
XX	SCN2A; anticonvulsant; muscular-Gen.; neuroprotective; antiarrhythmic;		
XX	antimigraine; nootropic; antiparkinsonian; neuroleptic; triannalizer;		
XX	antidepressant; analgesic; nephrotoxic; antidiabetic; cytostatic;		
XX	diagnostic; anxiety disorder; major depressive disorder; epilepsy;		
XX	paralytic; hyperthermia; myasthenia gravis; heart arrhythmia; ataxia;		
XX	migraine; Alzheimer's disease; Parkinson's disease; cystic fibrosis; pain;		
XX	inflammation; polycystic kidney disease; phobia; schizophrenia;		
XX	neuropathic pain; hyperglycemia; hyperinsulinemia; sodium channel;		
XX	muteln.		
OS	Homo sapiens.		
OS	Synthetic.		
XX			

FH Key Location/Qualifiers
 FT Misc-difference 892 /label= V8921
 FT /note= "wild-type Val substituted with Ile"
 XX
 PN WO2005014863-A1.
 XX
 PD 17-FEB-2005.
 XX
 PF 06-AUG-2004; 2004WO-AU001051.
 XX
 PR 07-AUG-2003; 2003AU-00904154.
 XX
 PA (BION-) BIONOMICS LTD.
 XX
 PI Mulley JC, Harkin LA, Dibbens LM, Phillips HA, Heron SE;
 PI Berkovic SF, Scheffer IE, Davy A;
 XX WPI; 2005-195767/20.
 DR N-PSDB; ADY27076.
 DR
 XX
 PT Identifying subject predisposed to disorder associated with ion channel
 PT dysfunction, involves determining presence of specific mutation event in
 PT genes encoding ion channel subunits.
 XX
 PS Claim 20; SEQ ID NO 84; 347bp; English.
 XX
 CC This invention describes a novel method of identifying a subject
 CC predisposed to disorder associated with ion channel dysfunction
 CC comprising ascertaining whether at least one of the genes encoding ion
 CC channel subunits in the subject has undergone a mutation. The invention
 CC also describes 1) isolated nucleic acid molecules encoding an isolated
 CC polypeptide which is a mutant or variant ion channel subunit (including a
 CC mutant KCNQ2 subunit, where the mutation event has occurred in C terminal
 CC domain of the subunit and leads to disturbance in the calmodulin binding
 CC affinity of the subunit) and 2) an expression vector, cells, antibodies
 CC and a method for producing a non-human transgenic animal which are all
 CC used for screening of candidate pharmaceutical agents for diagnosing or
 CC treating epilepsy or a disorder associated with ion channel dysfunction.
 CC The mutation detected in the method disrupts the functioning of an
 CC assembled ion channel so as to produce an epilepsy phenotype in the
 CC subject or produces one or more disorders associated with ion channel
 CC dysfunction such as hyper- or hypo-kalemic periodic paralysis, myotonias,
 CC malignant hyperthermia, myasthenia, cardiac arrhythmias, episodic ataxia,
 CC migraine, Alzheimer's disease, Parkinson's disease, schizophrenia,
 CC anxiety, depression, phobic obsessive symptoms, neuropathic pain,
 CC inflammatory pain, chronic/acute pain, Bartter's syndrome, polycystic
 CC kidney disease, Dent's disease, hyperinsulinemic hypoglycemia of infancy,
 CC cystic fibrosis, congenital stationary night blindness and total color-
 CC blindness in the subject or to produce an epilepsy phenotype when
 CC expressed in combination with one or more additional mutations or
 CC variations in the ion channel subunit genes. The products of the
 CC invention have anticonvulsant, muscular-Gen., neuroprotective,
 CC antiarrhythmic, antimigraine, nootropic, antiparkinsonian, neuroleptic,
 CC tranquilizer, antidepressant, analgesic, nephroprotective, antidiabetic and
 CC cytoskeletal activity. This sequence represents a fragment of the human
 CC sodium ion channel subunit SCN2A encoded by exon 16 which contains the
 CC mutation V8921.
 CC
 XX
 SQ Sequence 2005 AA;
 Query Match 61.0%; Score 6394.5; DB 9; Length 2005;
 Best Local Similarity 62.1%; Pred. No. 0;
 Matches 1300; Conservative 233; Mismatches 359; Indels 201; Gaps 33;
 QY 5 LIPRGSSFRFRTRSLAIEKRMKQAGSTTLOESREGLPDEEAPRPODLQSKYL 64
 DB 6 LVPPEGDSFRFRTRSLAIEKRIAEKAKRP---KQERKDDDDGCPGPNDSLDKGSJL 62
 QY 65 PDLGNPQOLIGEPLDLPFYSTQKTEIVLWKGKTIIRFSATNALVYLSPFHPIRRAA 124
 DB 63 PPIYGDIPPEMWSVPLEDDPYINKKTIIVLWKGKAIISRFATPALVILTFPNPIRKLA 122

QY 125 VKILVHSLFMNLMICTILITNCVFMADPPPEWTKYVETFTALITYTESLWKLARGCLH 184
 DB 123 IKILVHSLFMNLMICTILITNCVFMATNSNPDDMTKNVETFTGITYTFSLIKILARGCFLH 182
 QY 185 APTFLRDPNMWLDPSVILMAVTFEVDLGNVSAIRTRVIRALAKTISVIGLKTIVGALI 244
 DB 183 DFTLRDPNMWLDPSVILMAVTFEVDLGNVSAIRTRVIRALAKTISVIGLKTIVGALI 242
 QY 245 QSVKGLADVAVLVTFGLSVFALITGLQFMNIRHKCR-----NFTALNGTGSV 294
 DB 243 QSVKGLADVAVLVTFGLSVFALITGLQFMNIRHKCR-----NFTALNGTGSV 300
 QY 295 EADGLVME-----SLDLYLSDPENYLLKNGTSDVLICGNSSDAGTCPEGYRCIACAGENP 348
 DB 301 DNGTTRNRKRVISINMBEYIEDKSHFYFLGQNDALLCGNSSDAGTCPEGYRCIACAGENP 360
 QY 349 DHGYTSPDSPFAMAEIALFRIMTODCWERLYOQTIRSAKIYMFPMVLVIFLGSYYLVNLI 408
 DB 361 NYGTSFDTFEWAFLSFLRLMTQDFWENLYOLTIRAAKTYMIFPVLVIFLGSYYLVNLI 420
 QY 409 LAVVAMAYEBQNTTIAETEEKERPOEAMEMLKKEHALTIR-----GV 453
 DB 421 LAVVAMAYEBQNTTIAETEEKERPOEAMEMLKKEHALTIR-----GV 480
 QY 454 DTVSRSLIEMSPILAPVNSHE---RRSRKRKMSGTECEGEDRLPKSDSEDPGR----- 504
 DB 481 GVFESESSVASKLSKSEKELAKNRKKKKQKQEGGEE- KNDRLVKSSESDSIRKGRPF 539
 QY 505 -----MNNLSLITRGLSRTSMKRRSGSIPTFRRR- DLGSDADPADENS 549
 DB 540 SLEGSRLTYEKRFSPQSLISIRGSLFSPRRNRALFSPRGRAKIGSENDADEHS 599
 QY 550 TAGSESRHTSLV--WPLRRTAQQCPSPGCTA- IGHMLHGKNSVDCNGVSLIGA 606
 DB 600 TFEENDSRDLSLPVHRHGEHRHSNVSQASASRVLPLEPNMGHSVDCNGVSLVG- 658
 QY 607 GDPKATSPGSHLPVMLHPPDTTTPSEEGCPQMLTSQAPCYDGEEPGARALSAV 666
 DB 659 GPTSLTISAGOLL-----PGCTTTERI-RRRRSSHYVMDLLEDPISRQRAMSIA 708
 QY 667 SVLTSALEELSESRHKPCPCNNRLAQRVLYWECPLMWSIQGYKLYVMDPPTDITIMC 726
 DB 709 SILNTWELBELSEKQCPWCWYKANNCLIMDCCKPMLKXGLVNLVMDPFDVLAITIC 768
 QY 727 IVLNTLPAALHNYMTSFEEMLOVGNVFGIFTAETFTIILADPYVYQCGNNIPDS 786
 DB 769 IVLNTLPAALHNYMTSFEEMLOVGNVFGIFTAETFTIILADPYVYQCGNNIPDS 828
 QY 787 IIVILSMELGLSRMSNLVSRSFLLRVPFLAKSWPTLNTLIKIGNSVGALNTLVYL 846
 DB 829 FIVLSLMEGLANVEGLSVLRSPRLRVPFLAKSWPTLNTLIKIGNSVGALNTLVYL 888
 QY 847 AIIIVEIFAVVGMQIFGKNYS- LRSDSGLLPWHMMDFHAFLIRILICGEWIEIEMW 904
 DB 889 AIIIFIFAVVGMQIFGKNYS- LRSDSGLLPWHMMDFHAFLIRILICGEWIEIEMW 948
 QY 905 DCMETVSGSLCLVPLVLMVGNVNLFLAILLSSSAAVNLAPDREDRENNLQALATA 964
 DB 949 DCMETVSGSLCLVPLVLMVGNVNLFLAILLSSSAAVNLAPDREDRENNLQALATA 1008
 QY 965 RIQRGLRFRKRTTMDFCGGLRORPQKPAAL-----AAGQCPSCIAATPVSPPPETE 1017
 DB 1009 RMQKIDVVKAKIRF---IQKAPRKQKALDEIKPLEDLNKKDCSTIS----- 1054
 QY 1018 KVPPTRKETREEGE---OPGQGP-----GDPE-PYCVPIA 1050
 DB 1055 -----NHTTIEIGDLNLYLKDNGTGTGIGSSVEKYVVDSDYVSPINNLSLTVTVBIA 1108
 QY 1061 VASPTDQOEDEBENSLSCTEESSSQESQPVSGGEAPDPDRITMSQVSATPSSEAEASAS 1110
 DB 1109 VGEESDFE-----NNTTEEFSSSDME-----ESKELKATSSSEST--- 1145
 QY 1111 QADWRQKQKALEPQARCGCEPTEDSCSEGSTADMTNTAELLEQIPDLQGVDPEDCPTGEG 1170

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Db      1146 -----VDIGAPAEQGE-----VEPESELE-----PACFTED 1174
Qy      1171 CYRBCPCCAVDTTQAPGKWMMLRKTCHIVHSHFETITIMILLSSGALAFEDITYEE 1230
Db      1175 CYRKCRCOISIEEGKLMNMLRKYCYIVHSHFETITIMILLSSGALAFEDITYEO 1234
Qy      1231 RRTIVLEAYADKMFYFVLEMLKMWYGFKKFTNMCWLDPLIVSVSVANTL 1290
Db      1235 RRTITMELAYADKMFYFVLEMLKMWYGFKKFTNMCWLDPLIVSVSVANTL 1294
Qy      1291 GFAENGPIKSLRTLRALRPLRLASRPEGKRVVNVNLVCAIPSIMNVLVCLIFMILFSIM 1350
Db      1295 GYSEIGAIKSLRTLRALRPLRLASRPEGKRVVNVNLVCAIPSIMNVLVCLIFMILFSIM 1354
Qy      1351 GYNLFAGKFGRCINOTEGDPLNNTYTVNKSQCESL--NLTGELYMKVKNPDVNGAG 1407
Db      1355 GYNLFAGKFGRCINOTEGDPLNNTYTVNKSQCESL--NLTGELYMKVKNPDVNGAG 1411
Qy      1408 YVALLQVATFKGMMIMYAAVDSRGVEBQPOHEVNLVYVIFVIFIGSFETLNLFTGV 1467
Db      1412 YVLLQVATFKGMMIMYAAVDSRGVEBQPOHEVNLVYVIFVIFIGSFETLNLFTGV 1471
Qy      1468 IIDNFNOOKKKLGGODIFWTEBOKKYNNAMKKLGGKPOKPIPRPLNKYQGFIPDIWKO 1527
Db      1472 IIDNFNOOKKKLGGODIFWTEBOKKYNNAMKKLGGKPOKPIPRPLNKYQGFIPDIWKO 1531
Qy      1528 AFDVTIMFLICLNTVMTVETDQSEKINIILAKINLFLVALFTGECIVKALBRHYFT 1587
Db      1532 VEDISIMILICLNTVMTVETDQSEKINIILAKINLFLVALFTGECIVKALBRHYFT 1591
Qy      1588 NSMNIFFDPPVYVILSVGTSLDIIOKYFSPFLFRVIRARIGRIIRLRKAKGIRTLIF 1647
Db      1592 IGMNIFDFVYVILSVGTSLDIIOKYFSPFLFRVIRARIGRIIRLRKAKGIRTLIF 1651
Qy      1648 ALMMSLPALENIGLLFLVMPFYISFGMANFAYVYVKEADIDMPNFQFANMLCLFOIT 1707
Db      1652 ALMMSLPALENIGLLFLVMPFYISFGMANFAYVYVKEADIDMPNFQFANMLCLFOIT 1711
Qy      1708 TSAGMDGLSPILNTGPPYCDPTLPNSNGS-RDGCSPAVGILFFTYIIISFLIVNMY 1766
Db      1712 TSAGMDGLSPILNTGPPYCDPTLPNSNGS-RDGCSPAVGILFFTYIIISFLIVNMY 1771
Qy      1767 IAILIENSVAEESTEPSEDDFPMFYIRKPPKPPKATQFIRYSVLSFPALASEPLRI 1826
Db      1772 IAILIENSVAEESTEPSEDDFPMFYIRKPPKPPKATQFIRYSVLSFPALASEPLRI 1831
Qy      1827 AKPNQISILNMDLPMVSGDRICHMDILFAFKRVVGESEGMALKIOMEKEMANPSKI 1886
Db      1832 AKPNQISILNMDLPMVSGDRICHMDILFAFKRVVGESEGMALKIOMEKEMANPSKI 1891
Qy      1887 SYEPIITTLRRKHEBVSAMVIOARFRHLLQSLKHAFLFRQOAGSGISEBADEREGL 1946
Db      1892 SYEPIITTLRRKHEBVSAMVIOARFRHLLQSLKHAFLFRQOAGSGISEBADEREGL 1949
Qy      1947 IAYVNSNPSPRLGPSSSSISSTSPPSYDSVTRATSDNIOVRGSDVSHSD 1999
Db      1950 LIDKLNENST---PEKTDMTSTSPSPSYDSVTRATSDNIOVRGSDVSHSD 1995

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KW      diagnostic; anxiety disorder; major depressive disorder; epilepsy;
KW      paralysis; hyperthermia; amyotonia gravis; heart arrhythmia; ataxia;
KW      migraine; Alzheimer's disease; Parkinson's disease; cystic fibrosis; pain;
KW      inflammation; polycystic kidney disease; phobia; schizophrenia;
KW      neuropathic pain; hyperglycemia; hyperinsulinemia; sodium channel;
KW      mutin.
XX      Homo sapiens.
OS      Synthetic.
XX      Key
FH      Location/Qualifiers
FT      MISC-difference 1003
FT      /label= L1003I
FT      /note= "wild-type Leu substituted with Ile"
XX      WO2005014863-A1.
XX      17-FEB-2005.
XX      06-AUG-2004; 2004WO-AU001051.
XX      07-AUG-2003; 2003AU-00904154.
XX      (BION-) BIONOMICS LTD.
XX      Mulley JC, Harkin LA, Dibbens LM, Phillips HA, Heron SE;
XX      Berkovic SF, Scheffer IE, Davy A;
XX      WPI, 2005-195767/20.
XX      N-PSDB; ADY27077.
XX      Identifying subject predisposed to disorder associated with ion channel
XX      dysfunction, involves determining presence of specific mutation event in
XX      genes encoding ion channel subunits.
XX      Claim 20; SEQ ID NO 85; 347bp; English.
XX      This invention describes a novel method of identifying a subject
XX      predisposed to disorder associated with ion channel dysfunction
XX      comprising ascertaining whether at least one of the genes encoding ion
XX      channel subunits in the subject has undergone a mutation. The invention
XX      also describes 1) isolated nucleic acid molecules encoding an isolated
XX      polypeptide which is a mutant or variant ion channel subunit (including a
XX      mutant KCNQ2 subunit, where the mutation event has occurred in C terminal
XX      domain of the subunit and leads to disturbance in the calcium binding
XX      affinity of the subunit) and 2) an expression vector, cells, antibodies
XX      and a method for producing a non-human transgenic animal which are all
XX      used for screening of candidate pharmaceutical agents for diagnosing or
XX      treating epilepsy or a disorder associated with ion channel dysfunction.
XX      The mutation detected in the method disrupts the functioning of an
XX      assembled ion channel so as to produce an epilepsy phenotype in the
XX      subject or produces one or more disorders associated with ion channel
XX      dysfunction such as hyper- or hypo-kalemic periodic paralysis, myotonia,
XX      malignant hyperthermia, myasthenia, cardiac arrhythmias, episodic ataxia,
XX      migraine, Alzheimer's disease, Parkinson's disease, schizophrenia,
XX      anxiety, depression, phobic obsessive symptoms, neuropathic pain,
XX      inflammatory pain, chronic/acute pain, Bartter's syndrome, polycystic
XX      kidney disease, Dent's disease, hyperinsulinemic hypoglycemia of infancy,
XX      cystic fibrosis, congenital stationary night blindness and total color-
XX      blindness in the subject or to produce an epilepsy phenotype when
XX      expressed in combination with one or more additional mutations or
XX      variations in the ion channel subunit genes. The products of the
XX      invention have anticonvulsant, muscular-Gen., neuroprotective,
XX      antiarrhythmic, antimigraine, nootropic, antiparkinsonian, neuroleptic,
XX      tranquilizer, antidepressant, analgesic, nephrotoxic, antidiabetic and
XX      cytosolic activity. This sequence represents a fragment of the human
XX      sodium ion channel subunit SCN2A encoded by exon 17 which contains the
XX      mutation L1003I.
XX      Sequence 2005 AA;
XX      Query Match 61.0%; Score 6393.5; DB 9; Length 2005;
XX      Best Local Similarity 62.1%; Pred. No. 0;

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RESULT 27
 ADY27149
 ID ADY27149 standard; protein; 2005 AA.
 AC ADY27149;
 XX 05-MAY-2005 (first entry)
 DT
 XX Human SCN2A variant R223Q.
 XX
 KW SCN2A; anticonvulsant; muscular-Gen.; neuroprotective; antiarrhythmic;
 KW antimigraine; nootropic; antiparkinsonian; neuroleptic; tranquilizer;
 KW antidepressant; analgesic; nephrotoxic; antidiabetic; cytosolic;

XX AC AB883627;
 XX 10-OCT-2002 (first entry)
 XX DE Human GEFs+ protein with SCN2A mutation.
 XX KM Human, GEFs+, SCN2A, mutant, mutant, mutant;
 XX KM generalized epilepsy with febrile seizure plus.
 XX OS Homo sapiens.
 XX PN JP2002136289-A.
 PD 14-MAY-2002.
 PF 01-NOV-2000; 2000JP-00334969.
 PR 01-NOV-2000; 2000JP-00334969.
 PA (KAGA-) KAGAKU GIJUTSU SHINKO JIGYODAN.
 PA (RIKA) RIKAGAKU KENKYUSHO.
 DR MPI, 2002-552308/59.
 DR N-PSDB; AB079201.
 PT A human polynucleotide which is complementary to an mRNA transcribed from
 PT a generalized epilepsy with febrile seizure plus (GEFS+)-related gene
 PT useful for diagnosing GEFS+.
 PS Claim 10; Page 29-34; 37pp; Japanese.
 CC This invention relates to a human polynucleotide which is complementary
 CC to an mRNA transcribed from a "generalized epilepsy with febrile seizure
 CC plus" (GEFS+)-related gene. The gene is useful for diagnosing GEFS+. The
 CC present sequence represents the human GEFS+ protein sequence with SCN2A
 CC mutation
 XX Sequence 2005 AA;
 SQ

Query Match 60.9%; Score 6392.5; DB 5; Length 2005;
 Best Local Similarity 62.1%; Pred. No. 0;
 Matches 1300; Conservative 232; Mismatches 360; Indels 201; Gaps 33;

5 LIPRGTSFRRTRESLAIEKMAKQASSTLQESREGPEEAPRPOLDQASKL 64
 6 LVPPEPDSFFRFETRESLAIEQRIAEKAKRP---KQERKDEDENGEPNSDLEAGKSL 62
 65 PDLYGNPQELIGELDLDPFYSQKTFIVYINKGTFPRSATNALVLSFPHIRRAA 124
 63 PFYIGDIPPEWVSPELEDDPYINKTFIVYINKGKALSRSFATPALTLPFNIRKLA 122
 125 VKILVHSLENNLIMCTILTNCFMAQHPPTKTYVEYTFPAIYFESLVKILAGFCIH 184
 123 IKILVHSLENNLIMCTILTNCFMAQHPPTKTYVEYTFPAIYFESLVKILAGFCIE 182
 185 APTFLRDPWNLDFSVIIMAYTTEFVDLGNVSALFTFVRLAKTISVIGLKTIVGALI 244
 183 DFTFLRDPWNLDFSVIIMAYTTEFVDLGNVSALFTFVRLAKTISVIGLKTIVGALI 242
 245 OSVKKLADVMVLTVCCLSVFALIGLQFMGNLRHKCVR-----NFTALNTNSV 294
 243 OSVKKLADVMVLTVCCLSVFALIGLQFMGNLRHKCVR-----NFTALNTNSV 294
 295 EADGLVME-----SLDLYLSPENYLLKNGTSVLLCGNSSDAGTCPEGYRCLKAGEMP 348
 301 DNGGTTFRKTVSIFPMWDEYIEKSHFYFLEGONDALLCGNSDDAQCCEGYI CVYAKGNP 360
 349 DHGYSSTDSFAMAFIALFLMTQDCWERYQGTLSAGKIYIFPMVLIPLGSPYLVNLI 408
 361 NYGYSSTDSFAMAFIALFLMTQDCWERYQGTLSAGKIYIFPMVLIPLGSPYLVNLI 420
 409 LAVVAMAEQONQATIAETBEKRFQAMENLKKHEALITR-----GV 453

421 LAVVAMAEQONQATIAETBEKRFQAMENLKKHEALITR-----GV 453
 421 LAVVAMAEQONQATIAETBEKRFQAMENLKKHEALITR-----GV 453
 454 DTVSRSLSLEMSPLAPVNSHE--RRSKRKKNSSGTEBCEDRLPKSDESGPR----- 504
 481 GVFSSESSSVASKLSSKSEKELKNRRKKKKQEQSGEEE--KNDRVAKSESEDSIRKGRFR 539
 505 -----AMNHLSLRGLSRTSMKRRSSGSLFTRRR--DLSSEADPFADDS 549
 540 SLEGSRLTYEKRFSSPHOSLSIRGSLFPRNSASLPSFRGAKDIDGSEDPADDS 599
 550 TAGESHSRTSLVY--WPLRTSACQSPSEPTSA--PGHALGKNSTVDCGVVSLGA 606
 600 TFEEDNSRDLPLFPHRGERHNSVQASASRLPLIPNNGKSHASVDCGVVSLVG-- 658
 607 GDPKATSPGSHLRVPMLEHPDPTTPEEPGPGQMLTSQAPCVDFEPEGARQALSAV 666
 659 GPSTLTSAGQL-----PEGTTETEI--RRKRSSTHYHSMDLDEPDSRQAMSLA 708
 667 SVLTSALBELFESRHKPCPCNNRLAQRVLIWECCLMMSIQGVLYVMDPFTDLTIMC 726
 709 SILNTMELEESRQKPCPCNNRLAQRVLIWECCLMMSIQGVLYVMDPFTDLTIMC 768
 727 IVLNTLPMALBHYNNMTSEFEEMLOVGNLFTGIFTAEMTFKILADPPYFPOQGNIRDS 786
 769 IVLNTLPMALBHYNNMTSEFEEMLOVGNLFTGIFTAEMTFKILADPPYFPOQGNIRDS 828
 787 IIVLISLMEGLSRMSNLVLSRFLRLRVFKLANSWPTLNTLKIIGNSVGLAGNLTVL 846
 829 FIVLSLMEGLSRMSNLVLSRFLRLRVFKLANSWPTLNTLKIIGNSVGLAGNLTVL 888
 847 ALIYFIPAVNGMOLFGKRYSE--LRDSGSLPRHNMDFHAPLIRIIGENIETMW 904
 889 ALIYFIPAVNGMOLFGKRYSE--LRDSGSLPRHNMDFHAPLIRIIGENIETMW 948
 905 DCMESVGSGLCLVFLVAVGNLVNLFALILSFSADNLNAPDRRENNQLOLA 964
 949 DCMESVGSGLCLVFLVAVGNLVNLFALILSFSADNLNAPDRRENNQLOLA 1008
 965 RIORGARPKRTTWDPCGGLLRORPQKPAAL-----MAQGLSCIAATPSPPEPTE 1017
 1009 RMQGGIDPVRKIRBF--IKAFVRKQKALDEIKPLEDLNNKDCSCIS----- 1054
 1018 KVPFRKRTREBE-----OPGQSTP-----GDPE--PYCVPIA 1050
 1055 -----NHTTIEIKDNLVLDNGGTTSGISSVEKYVDSDDYMSFTNNSLTVTVPJA 1108
 1051 VASPTDQDEBDENSLGTRESESSKOSQOPVSGPEAPDSRTWSOVATSSSEAEASAS 1110
 1109 VGSODFE-----NLTTEFSSESDME-----ESKEKLNATSSSEGST--- 1145
 1111 QADRWQKAPQAPGCGETPDESCSEGSTADMTNLAELBOIDPQODVDPEDCFTEG 1170
 1146 -----VDIGAPABGEQPE-----VEPESSL-----PACCFED 1174
 1171 CVRRCPCCAVDTTQAPGKVMWRRLKTYHYHESWFEFIIIFMILSSGALAFEDYIEE 1230
 1175 CVRRCPCCAVDTTQAPGKVMWRRLKTYHYHESWFEFIIIFMILSSGALAFEDYIEE 1234
 1231 RKTIVLEVADKMFYVYVEMLLKNVAYGFKKYFTNACWMLDPLIYDVSIVSVANTL 1290
 1235 RKTIVLEVADKMFYVYVEMLLKNVAYGFKKYFTNACWMLDPLIYDVSIVSVANTL 1294
 1291 GFAEMGPIKSLRTLRALRPLRALSFRGMRVVANALVGALPSINNVLLVCLIFMLISIM 1350
 1295 GYSELGAIKSLRTLRALRPLRALSFRGMRVVANALVGALPSINNVLLVCLIFMLISIM 1354
 1351 GVNIPAGKFGKCIQTEGDLPLANTTYNNKQCSL--NLTSGLYTKVYVNDVNGAG 1407
 1355 GVNIPAGKFGKCIQTEGDLPLANTTYNNKQCSL--NLTSGLYTKVYVNDVNGAG 1411
 1408 YLALLQVATFKGMDIIVAAVDSRGYEQPOMENLVVYIFVFIIGSFPTNLFLIGV 1467

DR WPI, 2003-239332/23.
 DR N-PSDB; ADB78642.
 XX Identifying predisposition to an ion channel dysfunction, such as
 PT periodic paralysis, cardiac arrhythmias, migraine, Alzheimer's disease,
 PT schizophrenia, anxiety and depression, by detecting encoding-gene
 PT mutation events.
 XX
 PS Claim 13; SEQ ID NO 147; 106pp; English.
 XX
 CC The invention relates to a novel method for identifying a subject
 CC predisposed to a disorder associated with ion channel dysfunction. The
 CC method comprises ascertaining if at least one of the genes encoding ion
 CC channel subunits (ICS) has undergone a mutation event so that a cDNA
 CC derived from the subject has any of 134 nucleotide sequences. The method
 CC of the invention has nootropic, neuroprotective, inotropic, antipyretic,
 CC antiarthritic, antimitigative, antidepressant, antiparkinsonian,
 CC antileptemic, tranquilizer, analgesic, nephrotoxic, antidiabetic, and
 CC ophthalmological activity. A polynucleotide of the invention acts as an
 CC ion channel agonist, or ion channel antagonist. The methods, isolated
 CC nucleic acids, polypeptides, antibody, selective agonist, antagonist or
 CC modulator of an ion channel, cells and genetically modified non-human
 CC animal, are useful for the diagnosis and treatment of epilepsy and/or a
 CC disorder associated with ion channel dysfunction, such as hyper- or hypo-
 CC kalemia, periodic paralysis, myotonia, malignant hyperthermia,
 CC myasthenia, cardiac arrhythmias, episodic ataxia, migraine, Alzheimer's
 CC disease, Parkinson's disease, schizophrenia, hyperplexia, anxiety,
 CC depression, phobic obsessive symptoms, neuropathic pain, inflammatory
 CC pain, chronic/acute pain, Bartter's syndrome, polycystic kidney disease,
 CC Dent's disease, hyperinsulinaemic hypoglycaemia of infancy, cystic
 CC fibrosis, congenital stationary night blindness and total colour
 CC blindness. The present sequence represents a mutant protein of the
 CC invention. The sequence data for this patent is not represented in the
 CC printed specification, but was published in electronic format directly
 CC from WIPO at ftp.wipo.int/pub/published_pat_sequences.
 CC
 XX
 SO Sequence 2005 AA:
 Query Match 60.9%; Score 6391.5; DB 7; Length 2005;
 Best Local Similarity 62.1%; Pred. No. 0;
 Matches 1300; Conservative 232; Mismatches 360; Indels 201; Gaps 33;
 QY 5 LPRGSSRRFRFRESLAIEKRMKOKARGSTTLOESREG,PEEAPAPODLOASKU, 64
 DB 6 LVPBGDSFRFFRRESLAIEKRIEAKRP---KQERKDEDDGEPNSDLREGSL 62
 QY 65 PDLGNPPOELIGEPLEDLPPEYSTOKTEIVLNKGTIFRFSATNALVYLSPPHIRRA 124
 DB 63 PPIYGDIPPEWVSVPLEDLPYINKKTFIVLNKGAISRFSATPALVILTFPPIRKA 122
 QY 125 VKILVHSLNMLIMCTIILNCPMAOHPPEWTKYETFTALYTPESLVKILAGFCLH 184
 DB 123 IKILVHSLNMLIMCTIILNCPMTSNPPDWTKVNEYFTGTIYTESIKILARFCLE 182
 QY 185 APTFLRDPNNMIDESYIMAYTTEFYDILGNVSLRFTFVLRLAKTISVSGKTIYGAII 244
 DB 183 DTFLEDPNNMIDFVITFAYTTEFYDILGNVSLRFTFVLRLAKTISVSGKTIYGAII 242
 QY 245 QSVKSLADVWVLFVCLSVFALIGQLFMGNLEHKVCV-----NFTALNGTNGSV 294
 DB 243 QSVKSLADVWVLFVCLSVFALIGQLFMGNLEHKVCV-----NFTALNGTNGSV 300
 QY 295 EADGLWVE-----SLDLYISDPENYLNKNGTSVLLCGNSDPAGTCPEGTRCLAKGENP 348
 DB 301 DNGGTFRNFTVSIENNDVEYIEDKSHFYFLEGQNDALLCGNSDPAGTCPEGTRCLAKGENP 360
 QY 349 DHGYSFSDSPAFMFLFRIMTODCWERLYOQTLRSAGKIYMFMLVIFLGSFYVNI 408
 DB 361 NGYSFSDSPAFMFLFRIMTODCWERLYOQTLRSAGKIYMFMLVIFLGSFYVNI 420
 QY 409 LAVVANAAYEONQATTAEETEEKERFOEAMEMLKKEHEALTTR-----GV 453
 DB 421 LAVVANAAYEONQATTAEETEEKERFOEAMEMLKKEHEALTTR-----GV 480

QY 454 PTVRSLSLEMSPLAPVNSHE---RRSKRRKMSGTECEGDRLPKSDSEGGP----- 504
 DB 481 GVSESSSVASKLSKSEKELKRRKKKKQKQKQSGEE- KNDVLSKSESDDSIKRGFRF 539
 QY 505 -----AMNLSLTRGLSRTSMKPRSSRGSIFTFRRR--DLGSDPADDENS 549
 DB 540 SLEGSRLTYEKRFSSPHQSLSTRGSLFSPRRNSRALSFGRKADIGSENFADDEHS 599
 QY 550 TAGSESHRSLVLP--WPLRRTSAGQBPBGISA--RGMLHGKNSTVQCNVGSLLGA 606
 DB 600 TFEENDSRRLSLFVPHRHGERRSHNSVQASBASVLPILPMNGMBSAVDCNGVSLVG- 658
 QY 607 GDPATSPGSHLAPWMLHPDPTTPESEBGPQMLTSPAFCVGESEBQARORALSAV 666
 DB 659 GPSTLTSAGQL-----PEGTTTETEI--RRRSSYHVSMDLEDPTSRQANSLA 708
 QY 667 SVLTSALEBLESNHHKPCPCWNRRLAORVLIWECPLMWSIKQVLYVMDPFTDLTTMC 726
 DB 709 SILNTMELEBESRQKPCPCWYKFPANMCLWDCKPMLKVHVLNVLVMDPFDVLAITIC 768
 QY 727 IVANTLPMALRHNTSBEFEMLOVGNLVFTGIFTAEMTFKIILADPYVFOGQWNI 786
 DB 769 IVANTLPMALRHNTSBEFEMLOVGNLVFTGIFTAEMTFKIILADPYVFOGQWNI 828
 QY 787 IIVILSLMEIGLSHMSNLVLSRFLRVFLAKSNPTLNTLKIIGNSVAGALNLTVL 846
 DB 829 FIVLSLMEIGLSHMSNLVLSRFLRVFLAKSNPTLNTLKIIGNSVAGALNLTVL 888
 QY 847 AIIVFIPAVVGMOLFGKXYS--LRDSGSLPWRHMDPFHAFILIFRILGEMITMW 904
 DB 889 AIIVFIPAVVGMOLFGKXYS--LRDSGSLPWRHMDPFHAFILIFRILGEMITMW 948
 QY 905 DQMSVSGSLVLYELVMTVGNVNLFLALLSSPSADNLTAPEDRSMNLQALAA 964
 DB 949 DQMSVSGSLVLYELVMTVGNVNLFLALLSSPSADNLTAPEDRSMNLQALAA 1008
 QY 965 RIQGLARFVKRTTWDPCGGLRORPOKRAL-----AAQGLPSCIATPYSPPEPTE 1017
 DB 1009 RMQKIDIVKRIKEF---IQKAVRQKXALDEIKPLEDANNNKSDCS----- 1054
 QY 1018 KVPTRKREBGE---OPGQGP-----GDE- PUCVPIA 1050
 DB 1055 -----NNTTLEIGDNLVLTLDGNGTTSIGSSVEKYVDSDYMSFINNLSLTFTVPIA 1108
 QY 1061 VASPTDQDEEDENSLSCTEESSSKQESQPVSGGPBPDPRTMSQVSATPSSEASAS 1110
 DB 1109 VGESDPE-----NMTBEFSSSDME-----ESKELNATSSSGST-- 1145
 QY 1111 QADNRQKAEPOAPGCGETPEDESCSEGSTADMTNTAELLEQIDQGDVDPEDCFTEG 1170
 DB 1146 -----VIGAPABEGBE-----VEPESLSL-----PEACFTEG 1174
 QY 1171 CVRRPCCAVDTTQAPGVWNRRLKTYCIYVESHMFETFIIFMLSSGALAFEDYIEE 1230
 DB 1175 CVRRPCCAVDTTQAPGVWNRRLKTYCIYVESHMFETFIIFMLSSGALAFEDYIEE 1234
 QY 1231 RKTIVLLEFYADKMTYFVLEMLKXVAYGPKKYFTTNKCMDELIVDSVLSTVANTL 1290
 DB 1235 RKTIVLLEFYADKMTYFVLEMLKXVAYGPKKYFTTNKCMDELIVDSVLSTVANTL 1294
 QY 1291 GFAEWGPIKSLRTTALRPLRLSRFBGMRVVNVNLAISIMNVLLVLCIIFMLIFSIM 1350
 DB 1295 GFAEWGPIKSLRTTALRPLRLSRFBGMRVVNVNLAISIMNVLLVLCIIFMLIFSIM 1354
 QY 1351 GVNLPAGKRGRCINOTEGDLPANTYIVNKSQCSL---NLTGELYMTKXVNFQNVGAG 1407
 DB 1355 GVNLPAGKRGRCINOTEGDLPANTYIVNKSQCSL---NLTGELYMTKXVNFQNVGAG 1411
 QY 1408 YVALLQVATFKGMDIMYAAVDSKGYEQPOPMYVLYFVFIITIGSPFTLNLFTGV 1467
 DB 1412 YVALLQVATFKGMDIMYAAVDSKGYEQPOPMYVLYFVFIITIGSPFTLNLFTGV 1471

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QY 1468 IIDNPNQKKKGGGDI FMTBQKRYNNAMKLGSKKKPKPIRPLNKYGFIEDIVTKQ 1527
DB 1472 IIDNPNQKKKGGGDI FMTBQKRYNNAMKLGSKKKPKPIRPLNKYGFIEDIVTKQ 1531
QY 1528 APVVTMTPLCLAMVMTMTDQSPKINILAKINLPAITGGECIVKLAALRHYYFT 1587
DB 1532 VPDISIMILCLAMVMTMTDQSPKINILAKINLPAITGGECIVKLAALRHYYFT 1591
QY 1588 NSNNIDPFVVILISITGVLSIDIIOKYFSPPLFRVIRLARIRIRILRGAGIRTLIF 1647
DB 1592 IGMNIDPFVVILISITGVLSIDIIOKYFSPPLFRVIRLARIRIRILRGAGIRTLIF 1651
QY 1648 ALMMSLPALFNIGLILFLVMTFYSIRGMANFAVYKWEAGIDMFNFOTFANSMCLFQIT 1707
DB 1652 ALMMSLPALFNIGLILFLVMTFYSIRGMANFAVYKWEAGIDMFNFOTFANSMCLFQIT 1711
QY 1708 TSAQMDGLSPIANTGPPYCDPPLPNSNGS -RDCGSPAVGILFETTYIIISFLIVNMY 1766
DB 1712 TSAQMDGLSPIANTGPPYCDPPLPNSNGS -RDCGSPAVGILFETTYIIISFLIVNMY 1771
QY 1767 IAILNFSVATEESTEPLESDPDPMFYRIWEKDEDEATQFIEYSVLSDPADLSBPLRI 1826
DB 1772 IAILNFSVATEESTEPLESDPDPMFYRIWEKDEDEATQFIEYSVLSDPADLSBPLRI 1831
QY 1827 AKPNQISLIMDLPMTSGDRHCHMDILFAFTKRVLGESGEMDLAKIOMEKFFMAANPSKI 1886
DB 1832 AKPNQISLIMDLPMTSGDRHCHMDILFAFTKRVLGESGEMDLAKIOMEKFFMAANPSKI 1891
QY 1887 STEPIITTTLRKHEEVSAMVIOAFRRHLQSLKIASPLFRQAGSGISEDAPEREGL 1946
DB 1892 STEPIITTTLRKHEEVSAMVIOAFRRHLQSLKIASPLFRQAGSGISEDAPEREGL 1949
QY 1947 IAVWSENFSPRPSSSSISSTSPSPYDSVTFRATSDNLQVRGSDYHSHEP 1999
DB 1950 IAVWSENFSPRPSSSSISSTSPSPYDSVTFRATSDNLQVRGSDYHSHEP 1999
RESULT 31
ADB78605
ID ADB78605 standard; protein; 2005 AA.
AC ADB78605;
DT 04-DEC-2003 (first entry)
XX
DE Human sodium channel subunit mutant SEQ ID NO:149.
XX
KW mutect; mutant; ion channel; ion channel subunit; ICS; nootropic;
KW neuroprotective; inotropic; antipyretic; antiarrhythmic; antimigraine;
KW antidepressant; antiparkinsonian; neuroleptic; tranquilliser; analgesic;
KW neurotropic; antidiabetic; ophthalmological; epilepsy;
KW ion channel dysfunction; human.
XX
OS Synthetic.
OS Homo sapiens.
XX
PN WO2003008574-A1.
XX
PD 30-JAN-2003.
XX
PF 08-JUL-2002; 2002WO-AU000910.
XX
PR 18-JUL-2001; 2001AU-00006452.
PR 05-MAR-2002; 2002AU-0000910.
PR 13-MAY-2002; 2002AU-00002292.
XX
PA (BION-/) BIONOMICS LTD.
PA (WALL-) WALLACE R W.
XX
PI Mulvey JC, Herkin LA, Dibbens LM, Phillips HA, Heron SE,
PI Berkovic SF, Scheffer IE,
XX
DR WPI; 2003-239332/23.

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DR N-PSDB; ADB78644.
XX Identifying predisposition to an ion channel dysfunction, such as
PT periodic paralysis, cardiac arrhythmias, migraine, Alzheimer's disease,
PT schizophrenia, anxiety and depression, by detecting encoding-gene
XX mutation events.
PS Claim 13; SEQ ID NO 149; 1066p; English.
XX
CC The invention relates to a novel method for identifying a subject
CC predisposed to a disorder associated with ion channel dysfunction. The
CC method comprises ascertaining if at least one of the genes encoding ion
CC channel subunits (ICS) has undergone a mutation event so that a cDNA
CC derived from the subject has any of 134 nucleotide sequences. The method
CC of the invention has nootropic, neuroprotective, inotropic, antipyretic,
CC antiarrhythmic, antitachycardic, antidepressant, antiparkinsonian,
CC neuroleptic, tranquiliser, analgesic, nephrotoxic, antidiabetic, and
CC ophthalmological activity. A polynucleotide of the invention acts as an
CC ion channel agonist, or ion channel antagonist. The methods, isolated
CC nucleic acids, polypeptides, antibody, selective agonist, antagonist or
CC modulator of an ion channel, cells and genetically modified non-human
CC animal, are useful for the diagnosis and treatment of epilepsy and/or a
CC disorder associated with ion channel dysfunction, such as hyper- or hypo-
CC kalemia, periodic paralysis, myotonia, malignant hyperthermia,
CC myasthenia, cardiac arrhythmias, episodic ataxia, migraine, Alzheimer's
CC disease, Parkinson's disease, schizophrenia, hyperplexia, anxiety,
CC depression, phobic obsessive symptoms, neuropathic pain, inflammatory
CC pain, chronic/acute pain, Bartter's syndrome, poly cystic kidney disease,
CC Dent's disease, hyperinulinemic hypoglycaemia of infancy, cystic
CC fibrosis, congenital stationary night blindness and total colour
CC blindness. The present sequence represents a mutant protein of the
CC invention. The sequence data for this patent is not represented in the
CC printed specification, but was obtained in electronic format directly
CC from WIPO at ftp.wipo.int/pub/published_pat_sequences.
XX
SQ Sequence 2005 AA;
Query Match 60.9%; Score 6391.5; DB 7; Length 2005;
Query Local Similarity 62.1%; Pred. No. 0;
Matches 1300; Conservative 232; Mismatches 360; Indels 201; Gaps 33;
QY 5 LIPRGTSFRFRFTRESLAIEKMAKQARGSTTQESREGHPREEAPRPQDLQASKTL 64
DB 6 LVPPGPDSPFRFTRESLAIEKMAKQARGSTTQESREGHPREEAPRPQDLQASKTL 62
QY 65 PDLGNPQELIGLEPLEDDPYSTQKTFIVLNKKTIFRSATNALYVLSFPHPIRRA 124
DB 63 PRYGDIPPEWVSVPLEDDPYINKTPIYLNKKAISRFSATPALYLTFFNPPIRKA 122
QY 125 VKIIVHSLFNMLIMCTIILTNCFMAQHDPPMTKRYVETFRATYFESVYKILARFCIL 184
DB 123 IKILVHSLFNMLIMCTIILTNCFMMSNPDDTKNVEYFGIYFESIKILARFCIL 182
QY 185 AFTPLRDPWNLDPFVSIIMAYTTEFVDLGNVSALRTFRILAKTISVIGKTIYVGLI 244
DB 183 DFTPLRDPWNLDPFVSIIMAYTTEFVDLGNVSALRTFRILAKTISVIGKTIYVGLI 242
QY 245 QSVKGLADVMVLTFCISVFPALIGIQLFMGNLRHKVR-----NFTALNGTNGSV 294
DB 243 QSVKGLADVMVLTFCISVFPALIGIQLFMGNLRHKVR-----NFTALNGTNGSV 300
QY 295 EADGLVME-----SLDIYLSDPENYLLKNGSDVLTGNSSDACTCEGYCLAKGENP 348
DB 301 DNGTTFNRTVSIFFWDEYIEDKSHFYPLEGQNDLALGNSSDAQCCEGYCLAKGENP 360
QY 349 DHGYSFDSFAMAFALFRLMTQDCWRLYOOTLSAGKIYVIFMLYIFLGSFVLNLI 408
DB 361 NYGYSFDSFAMAFALFRLMTQDCWRLYOOTLSAGKIYVIFMLYIFLGSFVLNLI 420
QY 409 LAVVMAVEEONQATIAETEKEKRFQEAEMELKXHEALYIR-----GV 453
DB 421 LAVVMAVEEONQATIAETEKEKRFQEAEMELKXHEALYIR-----GV 480

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QY 454 DTVSRSLEWSPPLAVNSHE---RRGKRKRKMSGTEECGBDRLEPKSSENGPR----- 504
Db 481 GVFSSESSVASKLSKSEKELKNRRKKKKOKEOGEBEER-KQDRVLKSESESIIRKGRF 539
QY 505 -----ANNHSLTRGLSRTSMKPRSSGSIPTPRRR--DGSEADPADENS 549
Db 540 SLEGRLLYKRFKFSFHOSLSIRGSLSPRRNSRSLSPFRGRKADIGSEIDPADDHS 599
QY 550 TAGSESHRTSLV--WPLRRTSAOQOPSPTSA-PGHALHGKNSITVDCGVVSLGA 606
Db 600 TFEODNSRDLFLVHBRGERHSNVSOASRASRLPLPMNGKMSAIVDCGVVSLVG- 658
QY 607 GDEPATSPGSHLRVWMLHPRDPTTTPSEEPGPMLSQACVUGFEFGARQALSAV 666
Db 659 GPSTLTSAGQL-----PEGTTETEI--RKRRSSYHVSMDLSDPTSRQAMSAIA 708
QY 667 SVLTSALEBESRRKCPPCNRRLAORVLIWECPLMMSIKQGVKLVVMDPFTDLITMC 726
Db 709 SLTNTMBELBESRQKCPPCMYKAFANMCLIMDCCPKLVKVLVNLVMDPFDVLAITIC 768
QY 727 IYANTLFLMALHYNNTSEFEEMIQVGNLVTGIFTAEMTFKIIALDPYIYFOQWNIFDS 786
Db 769 IYANTLFLAMEMHYPTBEGSSVLASVGNLVTGIFTAEMFLKIIAMPYIYFOGWNIFDG 828
QY 787 IYIISLWELGSRMSNLSVLRSPFLRFLAKSPFTNTLITIGNSVGLAGNLTVL 846
Db 829 FIVSLISLWELGLANVGLSVLRSPFLRFLAKSPFTNTLITIGNSVGLAGNLTVL 888
QY 847 AIIVFIFAVVGMOLFQKNSY--LRSDSGLPRHMDPFPAFLIIFILGCEMIETMW 904
Db 889 AIIVFIFAVVGMOLFQKNSY--LRSDSGLPRHMDPFPAFLIIFILGCEMIETMW 948
QY 905 DCEWVSGOSLCLVFLVNVGNLVLVNLFLALLSSFSADNLAPDEDEKANNIQLALA 964
Db 949 DCEWVAGQIMCLTVPMVWVNLGNLVLVNLFLALLSSFSADNLAPDEDEKANNIQLAIVG 1008
QY 965 RIQRGLRFLYKRTTMDPCCGLRORPOKPAAL-----AAQQLPBCIATPPSPPEETE 1017
Db 1009 RMQKGDIFVKRKIRF---IQKAFVRKAKALDEIKLELDLNNKDCSIS----- 1054
QY 1018 KVPTRKETRFEEGE-----OPQGRP-----GDPE-PVCVPIA 1050
Db 1055 -----NNTTIELGKOLNTYKDGNGTTSIGSGSEVKYVDESVMYFINNPSLITVPIA 1108
QY 1051 VASDITDDEBEBENSAGTEBESSKQESQPVSGPEAPDPSRTSOSVATASSEASAS 1110
Db 1109 VGSDDP-----NLNTEEFSSSEDM-----ESKEKLANTSSESSEGT-- 1145
QY 1111 QADMROQKABPAPBCGCTPREDSCSEGSTADMNTAELLEQIPDLGQVQDPEDCFTG 1170
Db 1146 -----VDIGAPAEGEQPE-----VEBESLE-----PEACFTED 1174
QY 1171 CVRRCPCCAVDTTQAQKVMWRLRKTGYHIVESHMFETFIIFMILSSGALAFEDIYEE 1230
Db 1175 CVRRCPCCAVDTTQAQKVMWRLRKTGYHIVESHMFETFIIFMILSSGALAFEDIYEE 1234
QY 1231 RKTIKVLEBYADKFTYFVLEMLKMAVAGFKKYFTNACWLDPLIIVDSVLSVANTL 1290
Db 1235 RKTIKVLEBYADKFTYFVLEMLKMAVAGFKKYFTNACWLDPLIIVDSVLSVANTL 1294
QY 1291 GPEAMPISKLRTLRALRPLRALSREGMAYVNVNVALGALPSIMNVLVCLIFWLSFIM 1350
Db 1295 GPEAMPISKLRTLRALRPLRALSREGMAYVNVNVALGALPSIMNVLVCLIFWLSFIM 1354
QY 1351 GVNLPAGKPGRCINOTEGDPLNYTIVNKSOCESL---NLGELWYTKVAVFNDVAG 1407
Db 1355 GVNLPAGKPGRCINOTEGDPLNYTIVNKSOCESL---NLGELWYTKVAVFNDVAG 1411
QY 1408 YLALLQVATPKGMNDIMYAAVDSRGVEBQPMQENLWYIYFVIFIIFGSFFTLNPTGV 1467
Db 1412 YLALLQVATPKGMNDIMYAAVDSRGVEBQPMQENLWYIYFVIFIIFGSFFTLNPTGV 1471
QY 1468 IINFNQKKKKGGQDIFMTBQKKYNNAMKLGSKKPPQPIRPLNKYGPIFDIVTKQ 1527

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Db 1472 IINFNQKKKKGGQDIFMTBQKKYNNAMKLGSKKPPQPIRPLNKYGPIFDIVTKQ 1531
QY 1528 AFDVTIMFLCLNNVTMMVETDDOSPEKINIILAKINILFPAIFPGEIVLAAARHYFT 1587
Db 1532 VFDISIMILLCLNNVTMMVETDDOSPEKINIILAKINILFPAIFPGEIVLAAARHYFT 1591
QY 1588 NSNNIPFVVVYIISVGVVLSDIQKFFSPFLFRVIRLARIGILRLIRGAKGIRTLF 1647
Db 1592 IGMNIPFVVVYIISVGVVLSDIQKFFSPFLFRVIRLARIGILRLIRGAKGIRTLF 1651
QY 1648 ALMMSLPALFNIGLFLVMEIYISIFGMANFAYVKEAGIDMDMNFOTPANSMLCLFOIT 1707
Db 1652 ALMMSLPALFNIGLFLVMEIYISIFGMANFAYVKEAGIDMDMNFOTPANSMLCLFOIT 1711
QY 1708 TSAGMDGLSLPIANTGPPYCDPTLPNSNGS-RGQCGSPANGIIFETTYIIISFLVNMV 1766
Db 1712 TSAGMDGLSLPIANTGPPYCDPTLPNSNGS-RGQCGSPANGIIFETTYIIISFLVNMV 1771
QY 1767 IAILNENSVATERSTELSDDFDMFYEIWEKFPDEATOPFIEYSVLSDPADLSEPLRI 1826
Db 1772 IAILNENSVATERSTELSDDFDMFYEIWEKFPDEATOPFIEYSVLSDPADLSEPLRI 1831
QY 1827 AKPNOISLIMNDLPWVSGDRICHMDILFAFTKRVLGSGENDALKIQEKKFMAANPSKI 1886
Db 1832 AKPNOISLIMNDLPWVSGDRICHMDILFAFTKRVLGSGENDALKIQEKKFMAANPSKI 1891
QY 1887 SYEPTITTLRKAREVSAVMTQRAFRRLRLORSLKHSFLRQOAGSLSREDAPERGL 1946
Db 1892 SYEPTITTLRKAREVSAVMTQRAFRRLRLORSLKHSFLRQOAGSLSREDAPERGL 1949
QY 1947 IAYVSENFSPRLGPPSSSISSTSPSPSYSTRATSDNIQVGSYSHSED 1999
Db 1950 IAYVSENFSPRLGPPSSSISSTSPSPSYSTRATSDNIQVGSYSHSED 1999

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RESULT 32
ADCC46947
ID ADCC46947 standard; protein; 2005 AA.

AC ADCC46947;
XX
DT 18-DEC-2003 (first entry)
XX
DE Human SCN2A amino acid sequence #SEQ ID 3.
XX
KW SCN2A; voltage-gated ion channel; human; neuroprotective; gene therapy;
XX
OS Homo sapiens.
XX
PN WO2003060525-A1.
XX
PD 24-JUL-2003.
XX
PE 16-JAN-2003; 2003WO-EP000400.
XX
PR 17-JAN-2002; 2002EP-00001236.
XX
PR 17-JAN-2002; 2002US-0348674P.
XX
PA (EVOT-) EVOTEC NEUROSCIENCES GMBH.
XX
PI Hipfel R, Von Der Kammer H, Pohlner J;
XX
XX WPI; 2003-598580/56.
XX
XX DR N-PSDB; ADCC46947.
XX
XX PT Diagnosing or prognosticating a neurodegenerative disease by detecting
XX the level or activity of transcription or translation products of the
XX gene coding for the voltage-gated ion channel SCN2A.
XX
XX PS Disclosure; Fig 9; 67pp; English.
XX

CC The invention relates to a method for diagnosing or prognosticating a
 CC neurodegenerative disease in a subject, or determining whether a subject
 CC is at increased risk of developing the disease. The method comprises
 CC detecting the level and/or activity of a transcription or translation
 CC product of the gene coding for the voltage-gated ion channel SCN2A. The
 CC modulator of an activity and/or of a level of at least one substance is
 CC useful for preparing a composition for treating or preventing a
 CC neurodegenerative disease, in particular Alzheimer's disease. The current
 CC sequence represents the human SCN2A amino acid sequence.

XX
 CC
 Sequence 2005 AA:

Query Match 60.3%; Score 6391.5; DB 7; Length 2005;
 Best Local Similarity 62.1%; Pred. No. 0;
 Matches 1300; Conservative 232; Mismatches 360; Indels 201; Gaps 33;

QY 5 LIPRGSSRRFRRESLAIEKMAEKQARGSTTLOESRGLPEBEAPRQDLQASKL 64
 DB 6 LVPBGDSRFRFPRESLAIEQRIAEKAKRP--KQERKDDDENGPKNSDLKAGSL 62
 QY 65 PDLYGNPQELIGEPLEDPEYSTOKTFVLNKGKTIPEPSATNLVYSPHPIRBA 124
 DB 63 PFTYGIIPPEWASVPLEDDPYINKKTFVLNKGKAIISRFSPALPYLITPPNPIRKLA 122
 QY 125 VKLIWLSLFNMLIMCTILTNVCMADHPPEWTKYETFTATYTESLVKILARGFLH 184
 DB 123 IKLIWLSLFNMLIMCTILTNVCMADHPPEWTKYETFTATYTESLVKILARGFLH 182
 QY 185 APFFLDPMNMWLDPSYIIMAYTTEPVDLGNVSLRTFRVIRALKTTISVIGLKTIVGALI 244
 DB 183 DPFLLDPMNMWLDFTYITFAVYTFEVDLGNVSLRTFRVIRALKTTISVIGLKTIVGALI 242
 QY 245 QSVKRIADWVLTVPCLSVFALIGLQFMGNLPHKVR-----NFTALANGNSV 294
 DB 243 QSVKRIADWVLTVPCLSVFALIGLQFMGNLPHKVR-----NFTALANGNSV 294
 QY 295 EADGLVWE-----SIDLYLSPENYILNKGTSIDVLLCGNSDAGCCEGYRCLAKAGNP 348
 DB 301 DNGGTFRNRTVSIENMDEYIIBKSHFYLEGQNDALCGNSDAGCCEGYRCLAKAGNP 360
 QY 349 DHGITSFDSFANAFALFLMOTDCWERYLOOTLRSAKTIYMFMLVIFLAGFYLWNL 408
 DB 361 NGXYISFDFEFNAFSLFLMOTDFEMENYLOTLRAAGKTYMIFVLVIFLAGFYLWNL 420
 QY 409 LAVVMAAYEONATIAETEEKERFOZEMEMKKHEALTIR-----QY 453
 DB 421 LAVVMAAYEONATIAETEEKERFOZEMEMKKHEALTIR-----QY 453
 QY 454 DTYSRSSLEMSPLAPVNSHE---RSGRRKRMSSGTECGEDRLPKSDSEDPGR----- 504
 DB 481 GYFSESSVSAKLSSEKELKRRKKKQKQSGEEF-KNDRVYLSSESDSIRKGRFP 539
 QY 505 -----AMNHLSTRGSLSTSMKPRSSRGSITFRRR--DLGSEADPADENS 549
 DB 540 SLEGSRLTYEKFFSSPHQSLISIRGSLFSPRRNSRASLPSPFRARADIGSENDPADENS 599
 QY 550 TAGEBSHSTSLVLP--WPLRRTSAQGSFGTSA-PGHALGKKNSTYDCNGVSLTGA 606
 DB 600 TDEDNRDRLFPVRRHGERHSNVSQASRASRVLPILPNKGKMSAYDCNGVSLVVG- 658
 QY 607 GDPKATSPGSHLRPVMLEHPDPTTPPSRPGQPMVTSOAPCVDFEFGARORALSAV 666
 DB 659 GBSSTLSAGQL-----PGTTEETETI--RKRRSSSYHSMDLLEPTSRQRMSIA 708
 QY 667 SVLTSLALBELSRHKCPCCANRLAQRVLIWECCLPMSIKQGVKLVVMDPTDLTTINC 726
 DB 709 SLITVMEELSRHKCPCCANRLAQRVLIWECCLPMSIKQGVKLVVMDPTDLTTINC 768
 QY 727 IYALNTLPMALFHYNNMSEFEMLQVGNLVFTGIFTAEMTFKIIALDPYVYPOQGNITPS 786
 DB 769 IYALNTLPMALFHYNNMSEFEMLQVGNLVFTGIFTAEMTFKIIALDPYVYPOQGNITPS 828
 QY 787 IYVILSLMELGLSRMSNLVLRSPILRLVFKLAKSWPTLNTLTIKIIGNSVGLAGNLTVL 846

DB 829 FYISLSLMEGLANVGLSVLSPRLRLVFKLAKSWPTLNTLTIKIIGNSVGLAGNLTVL 888
 QY 847 AIIVFIYAVVGMQLPEKRYSE--LRSDSGLLPRHMHMPFFHFLIIFRLCGEWEITWM 904
 DB 889 AIIVFIYAVVGMQLPEKRYSE--LRSDSGLLPRHMHMPFFHFLIIFRLCGEWEITWM 948
 QY 905 DCMIEVSGSLICLLVFLVAVVIGLVNLFLALLSSFSADNLTAPDEDEKNNLQALALA 964
 DB 949 DCMIEVSGSLICLLVFLVAVVIGLVNLFLALLSSFSADNLTAPDEDEKNNLQALALA 1008
 QY 965 RIQGRLEFYKRTTWDFCCGLLRQPKPAL-----AAQQLPSCINATPYSPPEETE 1017
 DB 1009 RMQKGDIFVYKRIREFE---IQRAFYVKOKALDEIKPLEDLNNKDCIS----- 1054
 QY 1018 KVPFRKTERFEGE-----QCGQTP-----GDPE-PVCYPIA 1050
 DB 1055 -----NHTTIEIGKDLNLYLKQNGTTSIGISSEKRYVDESDYMSFINNPSLTVVPIA 1108
 QY 1051AVASDPTDQDEBENSIGTEESSKQESQPVSGPEAPDPSRTWSQVSAATASEASAS 1110
 DB 1109 VESDPE-----NLTDEFSSESDME-----ESKEKLNATSSSEGST--- 1145
 QY 1111 QADWROQWYABRQAPCEGETPEBDSCEGSTADMTNTAELLBOIPDLGQDVKPEDCFTEG 1170
 DB 1146 -----VDIGAPABEGQPE-----VEPEESLE-----PEACFTED 1174
 QY 1171 CYRRCPCCAVDTTQAPGKWMRLKTCYHIVHSNRETITFMILSSGALAEEDYLEE 1230
 DB 1175 CYRRCPCCAVDTTQAPGKWMRLKTCYHIVHSNRETITFMILSSGALAEEDYLEE 1234
 QY 1231 RRTIKVILEYADKMFYVVLVFLMLKVAVYGFKYFTNACMDPLIVDSVLSVANTL 1290
 DB 1235 RRTIKVILEYADKMFYVVLVFLMLKVAVYGFKYFTNACMDPLIVDSVLSVANTL 1294
 QY 1291 GRAEMGPISLRTLRALRPLRALSREGKRVVNVNVLGVAIPSIINVLVCLIFWILPSIM 1350
 DB 1295 GRAEMGPISLRTLRALRPLRALSREGKRVVNVNVLGVAIPSIINVLVCLIFWILPSIM 1354
 QY 1351 GYNLRAGKRCINOTBGDPLANYITVNKSGCESI---NLGELYTWYKYNFQVAVGAG 1407
 DB 1355 GYNLRAGKRCINOTBGDPLANYITVNKSGCESI---NLGELYTWYKYNFQVAVGAG 1411
 QY 1408 YLALLQVATEFKGMIMIMVAVDSRGYBQPOHEVNLVWYIVYVIFIIFGSPFTLNFIVG 1467
 DB 1412 YLALLQVATEFKGMIMIMVAVDSRGYBQPOHEVNLVWYIVYVIFIIFGSPFTLNFIVG 1471
 QY 1468 IIDNFQOQKKGQGDIFMTBEQKYYNNAMKLGSKKPKPIPRPLNKYOGFIFDVTXQ 1527
 DB 1472 IIDNFQOQKKGQGDIFMTBEQKYYNNAMKLGSKKPKPIPRPLNKYOGFIFDVTXQ 1531
 QY 1528 AFDVTIMFLICLNVMTVMVETDQSPKINIILAKINLFLVALFTGBCIVKLAALRHYFT 1587
 DB 1532 AFDVTIMFLICLNVMTVMVETDQSPKINIILAKINLFLVALFTGBCIVKLAALRHYFT 1591
 QY 1588 NSWNIIDFVAVVLSIVGTVLSIIQKYFSPFLFVIVIRARIGRIILRLRGAKGIFTLF 1647
 DB 1592 NSWNIIDFVAVVLSIVGTVLSIIQKYFSPFLFVIVIRARIGRIILRLRGAKGIFTLF 1651
 QY 1648 ALMMSLPALENIGLILFLVMFYYSIFGMANFAYVYKBEAGIDDMFNFQTPANMLCLFOIT 1707
 DB 1652 ALMMSLPALENIGLILFLVMFYYSIFGMANFAYVYKBEAGIDDMFNFQTPANMLCLFOIT 1711
 QY 1712 TSAGWDGLAPLANSRPPDCPDKHPGSSSVVGDGNSVGVGIFFEVSYIILIFLVLVNNY 1771
 DB 1767 TSAGWDGLAPLANSRPPDCPDKHPGSSSVVGDGNSVGVGIFFEVSYIILIFLVLVNNY 1826
 QY 1772 IAVILNENSVATSEASRPSDDFEMFYEWKDFPDATQIFEFKLSDFADLPPPLI 1831
 DB 1827 AKPNQISLINDLPMVSGDRICHMDILPAFTKRVVGESEMDALKIOMEKEMANPSKI 1886


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Db 1146 -----VDIGAPAGEQPE-----VEPESELE-----PEACPTED 1174
Qy 1171 CVRRCCCAVDTTQAPGKVMWRRLKTCYHIVESHWFETFIEMTILSSGALAEFDYLEE 1230
Db 1175 CVRRKCCQISIEGKLMWNRKTCYKVEHNPETFTVEMILSSGALAEBDYIEQ 1234
Qy 1231 RKTIKVLEAYDKMFTVVFVLEMLKVAAGFKKYPFNACMDPLIVDSLVSVANTL 1290
Db 1235 RKTIKMLEYADKVFYFIIFLEMLKVAAGFOYFFFNACMDPLIVDSLVSVANTL 1294
Qy 1291 GFAEMGPIKSLRTLRALRPLRALSREGMKRVVNAVGALPSIMNVLLVCLIFMILFSIM 1350
Db 1295 GYSELGAIKSLRTLRALRPLRALSREGMKRVVNAVGALPSIMNVLLVCLIFMILFSIM 1354
Qy 1351 GNVLPAGKRGRCINQEGDPLATYTVNKSQCESL---NLGELVWTKRVKVPDVGAG 1407
Db 1355 GNVLPAGKRGRCINQEGDPLATYTVNKSQCESL---NLGELVWTKRVKVPDVGAG 1411
Qy 1408 YLALQVATFKGMDIMYAADVSRGYEEOQPMQENLYMYFIYFIIFGSPFLNLEIGV 1467
Db 1412 YLSTLQVATFKGMDIMYAADVSRNVELQPKYEDNLYMYFIYFIIFGSPFLNLEIGV 1471
Qy 1468 IIDNFVQOKKKLGGQDIEMTEQOKKYNNAMKLGSKKPKQPIPRPANKYOGPIFDIVTKQ 1527
Db 1472 IIDNFVQOKKKLGGQDIEMTEQOKKYNNAMKLGSKKPKQPIPRPANKYOGPIFDIVTKQ 1531
Qy 1528 APDVIMELICLMMVTMMVETDQSPKINIILAKINLLPAITGECIVGLALRRHYPT 1587
Db 1532 VEPDITMILICLMMVTMMVETDQSPKINIILAKINLLPAITGECIVGLALRRHYPT 1591
Qy 1588 NSNNIDPFVNVILSIYGVTVLSIIQKFFSPTLFVRVRLARIRRLIRLKGAGIRTLAF 1647
Db 1592 IGNITDFVNVILSIYGVTVLSIIQKFFSPTLFVRVRLARIRRLIRLKGAGIRTLAF 1651
Qy 1648 ALMMSLPALFNIGLLFLVWFYISIFGMANFAYVKKWAGIDMNFQTFANSMCLFOIT 1707
Db 1652 ALMMSLPALFNIGLLFLVWFYISIFGMANFAYVKKWAGIDMNFQTFANSMCLFOIT 1711
Qy 1708 TSGMGGLSPIANTGPPYCDPLTPNSNGS-RDCCSPAVGLIFFTYIIISFLYVNMV 1766
Db 1712 TSGMGGLSPIANTGPPYCDPLTPNSNGS-RDCCSPAVGLIFFTYIIISFLYVNMV 1771
Qy 1767 IAIILNFVATSESTEPSDEDFDMFYETWEKDEDEAFOFIYSVLSPADLSPLRI 1826
Db 1772 IAIILNFVATSESTEPSDEDFDMFYETWEKDEDEAFOFIYSVLSPADLSPLRI 1831
Qy 1827 AKNQSLINMDLPMVSGDRHICMDLFAFTKRVLGESGEMDLAKTOMEKFFMAANPSKI 1886
Db 1832 AKNQSLINMDLPMVSGDRHICMDLFAFTKRVLGESGEMDLAKTOMEKFFMAANPSKI 1891
Qy 1887 STEPIITTLRKHEEVSAMVTOAPRRHLQSLKIASFLFRQAGSGISEDAPREGL 1946
Db 1892 STEPIITTLRKHEEVSAMVTOAPRRHLQSLKIASFLFRQAGSGISEDAPREGL 1949
Qy 1947 IAYVWSENSRPLPSPSSSISSTSPSYDSVTARNDMLQVRGSDYSHSD 1999
Db 1950 IAYVWSENSRPLPSPSSSISSTSPSYDSVTARNDMLQVRGSDYSHSD 1995

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KM paralytic; hyperthermia; myasthenia gravis; heart arrhythmia; ataxia;
KM migrating; Alzheimers disease; Parkinsons disease; cystic fibrosis; pain;
KM inflammation; polycystic kidney disease; phobia; schizophrenia;
KM neuropathic pain; hyperglycemia; hyperinsulinemia; sodium channel;
KM mutein.
OS Homo sapiens.
XX Synthetic.
FH Key Location/Qualifiers
FT MISC-difference 1319
FT /label= R1319Q
FT /note="wild-type Arg substituted with Gln"
XX
XX WO2005014863-A1.
XX
XX 17-FEB-2005.
XX
XX 06-AUG-2004; 2004WO-AU001051.
XX
XX 07-AUG-2003; 2003AU-00904154.
XX
XX (BION-) BIONOMICS LTD.
XX
XX Muller JC, Harkin LA, Dibbens LM, Phillips HA, Heron SB;
XX Berkovic SF, Scheffer IE, Davy A;
XX WPI, 2005-195767/20.
XX N-PSDB; ADY27079.
XX
XX PT Identifying subject predisposed to disorder associated with ion channel
XX PT dysfunction, involves determining presence of specific mutation event in
XX PT genes encoding ion channel subunits.
XX
XX Claim 20; SEQ ID NO 87; 347pp; English.
XX
XX This invention describes a novel method of identifying a subject
XX predisposed to disorder associated with ion channel dysfunction
XX comprising ascertaining whether at least one of the genes encoding ion
XX channel subunits in the subject has undergone a mutation. The invention
XX also describes 1) isolated nucleic acid molecules encoding an isolated
XX polypeptide which is a mutant or variant ion channel subunit (including a
XX mutant KCNQ2 subunit, where the mutation event has occurred in C terminal
XX domain of the subunit and leads to disturbance in the calmodulin binding
XX affinity of the subunit) and 2) an expression vector, cells, antibodies
XX and a method for producing a non-human transgenic animal which are all
XX used for screening of candidate pharmaceutical agents for diagnosing or
XX treating epilepsy or a disorder associated with ion channel dysfunction.
XX The mutation detected in the method disrupts the functioning of an
XX assembled ion channel so as to produce an epilepsy phenotype in the
XX subject or produces one or more disorders associated with ion channel
XX dysfunction such as hyper- or hypo-kalemic periodic paralysis, myotonia,
XX malignant hyperthermia, myasthenia, cardiac arrhythmias, episodic ataxia,
XX migraine, Alzheimer's disease, Parkinson's disease, schizophrenia,
XX anxiety, depression, phobic obsessive symptoms, neuropathic pain,
XX inflammatory pain, chronic/acute pain, Bartter's syndrome, polycystic
XX kidney disease, Dent's disease, hyperinsulinemic hypoglycemia of infancy,
XX cystic fibrosis, congenital stationary night blindness and total color-
XX blindness in the subject or to produce an epilepsy phenotype when
XX expressed in combination with one or more additional mutations or
XX variations in the ion channel subunit genes. The products of the
XX invention have anticonvulsant, muscular-Gen., neuroprotective,
XX antiarrhythmic, antidepressant, anxiolytic, antiparkinsonian, neuroleptic,
XX tranquilizer, antidepressant, analgesic, nephroprotective, antidiabetic and
XX cytoskeletal activity. This sequence represents a fragment of the human
XX sodium ion channel subunit SCN2A encoded by exon 20 which contains the
XX mutation R1319Q.
XX
XX Sequence 2005 AA;
XX
XX Query Match 60.9%; Score 6391.5; DB 9; Length 2005;
XX Best Local Similarity 62.1%; Pred. No. 0;
XX Matches 1300; Conservative 233; Mismatches 359; Indels 201; Gaps 33;

```

QY 5 LLPRGTSFRFRFTRESLAAIEKMAEKOARGSTTLQESREGLPEBEAPRQDLOQAKCL 64
 Db 6 LVPFPDPSFRFTRESLAAIEKRIAEKAKR---KQERKDEDDNGKRPMSDLAEGKSL 62
 QY 65 PDLVGNPQOELIGEPELDLPFYSTQKTFYVANKGKTTFRSATNALVLSFPHIRQAA 124
 Db 63 PFIYGDIPPEWVSVPLELDLPYINKKTFIYVANKKAISRFSATPALYILPFPNIRKLA 122
 QY 125 VKIIVHSLPNNMLINCTIILTNCEVMAOHDPBPWTKKVETFTAIYFEESI VKIILAGFCIH 184
 Db 123 IKIIVHSLPNNMLIKCTIILTNCEVPMWNSNPDMWKVETFTGITYFRESLIKILAGFCIE 182
 QY 185 AFTPLRDPNNMLDFSVIIMAYTTEPFVLCGNVSAKTRFVRLAKTISVIGLKTIVGALI 244
 Db 183 DTFPLRDPNNMLDFYITFAVYTEPFVLCGNVSAKTRFVRLAKTISVIGLKTIVGALI 242
 QY 245 QGVKKLADVMVLTVECLSVFALIGLQIFNGNLRHKCVR-----NFTLANGTNSV 294
 Db 243 QGVKKLADVMVLTVECLSVFALIGLQIFNGNLRHKCVR-----NFTLANGTNSV 300
 QY 295 KADGLVME-----SLDVLSDPENYLLKNGTSDVLLCGNSSDAGTCPEGRCLKAGEHP 348
 Db 301 DNGYTFNRTVSIENWDEYIEDSKHFYFLBEGNDALLCGNSSDAGTCPEGRCLKAGEHP 360
 QY 349 DHGYTSPDSFPAFAFLFRLMTQDCWERTYQOTLRSAKTIYIIFMLVIFLGSFYLVNLI 408
 Db 361 NGYTSFDTFSNAFLSLFRMTQDPFENLYQTLRAAGKTYMIFVLVIFLGSFYLVNLI 420
 QY 409 LAVVANAEBONQATIAETBEKKEKFOEAMEMLKKEHEALTR-----GV 453
 Db 421 LAVVANAEBONQATIEAEQKEAEFOQMLEKQOEEOAIAAAAAAASRDPSGAGI 480
 QY 454 DTVSRSLSPRLAPVNSH---RBRKRRKMSGTEEGEDRLPKSDESDGR----- 504
 Db 481 GVFESESSVASKLSSSEKELKNRRKKKQKEQSGEEB-KNDVLKSSBEDSIRKGRFP 539
 QY 505 -----ANMHLSTLRGLSRTSMKPRSSRSIIFTRRR--DLGSEADPADDENS 549
 Db 540 SLEGSRLTYEKRSSPHQSLSTRGSLFSPRRNSRSLSPFRGRADISENDPADDENS 599
 QY 550 TAGESBSHRTSLVP--WPLRRTSAQGPSPGTSA-PGHALHKKNSYDCCNGVSLLA 606
 Db 600 TFEFNDNSRRDLSLFVPHHGERHSHSNVQASRASRVPIILPMNKKMSAVDCCNGVSLV 658
 QY 607 GDEBARSPGSHLRPWLHPRPDTTPSEERPGPQWLTQAPCVDGFEERGARQRLASAV 666
 Db 659 GPSTLTSAQQL-----PEGTTTETET--RKRSSSYHVSMDLLEDTSKQRAMSTA 708
 QY 667 SVLTSALEBLESBRHKPCPCMNRLAQRLLIWECCPLWMSIKQGVKLVMDFPTDLTTWC 726
 Db 709 SILTNMEELESBRQKCPPCMYKPAWNCMLWDCKPKLVKGLVNLVWMDPFVDAITTC 768
 QY 727 IVANTLLEMALEHYNTSEFEEDMLQVGNLVTTGIFTAEMFKTIALDPYTFQGNKIPDS 786
 Db 769 IVANTLLEMALEHYNTSEFEEDMLQVGNLVTTGIFTAEMFKTIALDPYTFQGNKIPDS 828
 QY 787 IYVILSLMELGSLMSMLSVLRSFRLRVRKLAWSWTLTLTKIIGNSGALGNLTVL 846
 Db 829 FIVLSLMELEGLAVNEGSLVRSFRLRVRKLAWSWTLTLTKIIGNSGALGNLTVL 888
 QY 847 AIIVFIFAVVGMOLFQKNYSE--LRDSGGLPRMHMDFEHAFLIFRILICGMETW 904
 Db 889 AIIVFIFAVVGMOLFQKNYSE--LRDSGGLPRMHMDFEHAFLIFRILICGMETW 948
 QY 905 DCEVSGQSCLLAVLLVMVIGNLVNLFLALLLSSPSADNLTAPDEBERNNLQALAA 964
 Db 949 DCEVAGQTCWCLTFVMMVMVIGNLVNLFLALLLSSPSADNLTAPDEBERNNLQALAA 1008
 QY 965 RIQGLARFVKTTWDFCCGLLRQRPQRAL-----AAGQPLSCIATPYSPPETE 1017
 Db 1009 RMQKIDIFVKKRIREF--IKAFVRKQKALDEIKPLELDNNKDCSIS----- 1054

QY 1018 KVPETREKTEFESE-----QPGQGP-----GDPE-PVCVPRIA 1050
 Db 1055 -----NHTTILEGKOLANYLKQNGTTISGIGSSVEKRYVDESDYMRINNPSLTVTPRIA 1108
 QY 1051 VAESDTPDOEBESENSIGTEESSKQESQVSGQPEAPPDSTRWSQVATASSAEASAS 1110
 Db 1109 VGESDPE-----NLTTEFSSESDEME-----ESKEKLNATSSSEGS7-- 1145
 QY 1111 QADMROQKAKPQAPCGGEFPEBSCSBSGTADMTNTAELEQIPDLQGVKXDEPDCTEG 1170
 Db 1146 -----VDIGAPAGEQPE-----VEPESLE-----PEACTED 1174
 QY 1171 CVRRCPCCAVDTTQAPGVMMRLAKTCYHIVHSMFETFIIMILSSGALAFEDIYLB 1230
 Db 1175 CVRRFKCCQISIEBGKGLMMNLKCTCYKIVHNWFFTLFVFMILSSGALAFEDIYLB 1234
 QY 1231 RKTIKVLEVDKMFYVFLVLEMLLKVAVGFKYFTNACWDLFLVDVSLSVANTL 1290
 Db 1235 RKTIKVLEVDKMFYVFLVLEMLLKVAVGFKYFTNACWDLFLVDVSLSVANTL 1294
 QY 1291 GFAEMGPKISLRTLRALRPLALSFRGMRVYVNAALGALPSIMNVLLVCLIFMLIFSIM 1350
 Db 1295 GYSEIGAKISLRTLRALRPLALSFRGMRVYVNAALGALPSIMNVLLVCLIFMLIFSIM 1354
 QY 1351 GVNLPAGKEGRGINOTEGDPLANTYIVNKSQCESL---NLTGELVYTKVKNFNDYAG 1407
 Db 1355 GVNLPAGKEGRGINOTEGDPLANTYIVNKSQCESL---NLTGELVYTKVKNFNDYAG 1411
 QY 1408 YLALLOVATFKGMDIMYAADSRYEBOQMEYNLYIYFVIFIIFGSFPTNLPIGV 1467
 Db 1412 YLALLOVATFKGMDIMYAADSRYEBOQMEYNLYIYFVIFIIFGSFPTNLPIGV 1471
 QY 1468 IINDFNQOKKLGQGDIFMTSEOKKYIYAMKGLSKKRPQKTIIPPLNKYOFIFDIYTKQ 1527
 Db 1472 IINDFNQOKKLGQGDIFMTSEOKKYIYAMKGLSKKRPQKTIIPPLNKYOFIFDIYTKQ 1531
 QY 1528 AFDVTIMELICANWYTMVETDDOSPEKINTIILAKINILFVALIFGEICIVKLAALRHYYFT 1587
 Db 1532 VFDISIMELICANWYTMVETDDOSPEKINTIILAKINILFVALIFGEICIVKLAALRHYYFT 1591
 QY 1588 NSMNIFFDVFVYVILSVGTSLSDIIOKYPFSPFLFRVIRLARIIRILIRAKGIRTLF 1647
 Db 1592 IGMNIFDVFVYVILSVGTSLSDIIOKYPFSPFLFRVIRLARIIRILIRAKGIRTLF 1651
 QY 1648 ALMSGLPALFNIGLILFLVMFYISFGANPAAVYKMEGIDMFMFOFANSMCLFOIT 1707
 Db 1652 ALMSGLPALFNIGLILFLVMFYISFGANPAAVYKMEGIDMFMFOFANSMCLFOIT 1711
 QY 1708 TSAGMDGLSPILNTGPPYCDPTLPNSNGS--RGDCGSPAVGILFPTTYIIISFLIVNMY 1766
 Db 1712 TSAGMDGLSPILNTGPPYCDPTLPNSNGS--RGDCGSPAVGILFPTTYIIISFLIVNMY 1771
 QY 1767 IAIILLENFSVATESEPLESDDFMFEIWEKFPDEATOFIYESVLSDFADALSEPRLI 1826
 Db 1772 IAIILLENFSVATESEPLESDDFMFEIWEKFPDEATOFIYESVLSDFADALSEPRLI 1831
 QY 1827 AKPNQISLINDMLPMVSGDRHICMDILPAFTKRVYGESEGMALIKOMEKEMANPSKI 1886
 Db 1832 AKPNQISLINDMLPMVSGDRHICMDILPAFTKRVYGESEGMALIKOMEKEMANPSKI 1891
 QY 1887 GYEPITTTLRARHGEVSANVIOARFPHLOSRLKASPLFQOAGSGISEEDABERGL 1946
 Db 1892 GYEPITTTLRARHGEVSANVIOARFPHLOSRLKASPLFQOAGSGISEEDABERGL 1949
 QY 1947 IAYVSENFSPRLGPPSSSISSTSPSPSYDSVTATSDNLOVRSQDYSHSD 1999
 Db 1950 IADKANENST-----PEKTDMTSTTSPSPSYDSVTATSDNLOVRSQDYSHSD 1995

RESULT 35
 ADY27150
 ID ADY27150 standard; protein; 2005 AA.
 XX

AC ADY27150;
 XX 05-MAY-2005 (first entry)
 XX
 DE Human SCN2A variant R223Q.
 XX
 KM SCN2A; anticonvulsant; muscular-Gen.; neuroprotective; antiarrhythmic;
 KM antimigraine; nootropic; antiparkinsonian; neuroleptic; tranquillizer;
 KM antidepressant; analgesic; nephrotoxic; antidiabetic; cytostatic;
 KM diagnostic; anxiety disorder; major depressive disorder; epilepsy;
 KM paralytic; hyperthermia; myasthenia gravis; heart arrhythmia; ataxia;
 KM migraine; Alzheimer's disease; Parkinson's disease; cystic fibrosis; pain;
 KM inflammation; polycystic kidney disease; phobia; schizophrenia;
 KM neuropathic pain; hyperglycemia; hyperinsulinemia; sodium channel;
 KM mutein.
 XX
 OS Homo sapiens.
 OS Synthetic.
 XX
 FH Key Location/Qualifiers
 FT Misc-difference 1200 /label= T1200A
 FT /note= "wild-type Thr substituted with Ala"
 XX
 PN W02005014863-A1.
 XX
 PD 17-FEB-2005.
 XX
 XX 06-AUG-2004; 2004W0-AU001051.
 XX
 PR 07-AUG-2003; 2003AU-00904154.
 XX
 PA (BION-) BIONOMICS LTD.
 XX
 PI Mulley JC, Harkin LA, Dibbens LM, Phillips HA, Heron SE;
 PI Berkovic SF, Scheffer IE, Davy A;
 XX
 DR MPI; 2005-195767/20.
 DR N-PDB; ADY7078.
 XX
 PT Identifying subject predisposed to disorder associated with ion channel
 PT dysfunction, involves determining presence of specific mutation event in
 PT genes encoding ion channel subunits.
 XX
 PS Claim 20; SEQ ID NO 66; 347pp; English.
 XX
 CC This invention describes a novel method of identifying a subject
 CC predisposed to disorder associated with ion channel dysfunction
 CC comprising ascertaining whether at least one of the genes encoding ion
 CC channel subunits in the subject has undergone a mutation. The invention
 CC also describes 1) isolated nucleic acid molecules encoding an isolated
 CC polypeptide which is a mutant or variant ion channel subunit (including a
 CC mutant KCNQ2 subunit, where the mutation event has occurred in C terminal
 CC domain of the subunit and leads to disturbance in the calmodulin binding
 CC affinity of the subunit) and 2) an expression vector, cells, antibodies
 CC and a method for producing a non-human transgenic animal which are all
 CC used for screening of candidate pharmaceutical agents for diagnosing or
 CC treating epilepsy or a disorder associated with ion channel dysfunction.
 CC The mutation detected in the method disrupts the functioning of an
 CC assembled ion channel so as to produce an epilepsy phenotype in the
 CC subject or produces one or more disorders associated with ion channel
 CC dysfunction such as hyper- or hypo-kalemic periodic paralysis, myotonia,
 CC malignant hyperthermia, myasthenia, cardiac arrhythmias, episodic ataxia,
 CC migraine, Alzheimer's disease, Parkinson's disease, schizophrenia,
 CC anxiety, depression, phobic obsessive symptoms, neuropathic pain,
 CC inflammatory pain, chronic/acute pain, Bartter's syndrome, polycystic
 CC kidney disease, Dent's disease, hyperinsulinemic hypoglycemia of infancy,
 CC cystic fibrosis, congenital stationary night blindness and total color-
 CC blindness in the subject or to produce an epilepsy phenotype when
 CC expressed in combination with one or more additional mutations or
 CC variations in the ion channel subunit genes. The products of the
 CC invention have anticonvulsant, muscular-Gen., neuroprotective,
 CC antiarrhythmic, antimigraine, nootropic, antiparkinsonian, neuroleptic,

CC tranquillizer, antidepressant, analgesic, nephrotoxic, antidiabetic and
 CC cytostatic activity. This sequence represents a fragment of the human
 CC sodium ion channel subunit SCN2A encoded by exon 19 which contains the
 CC mutation T1200A.
 CC
 XX
 SQ Sequence 2005 AA;
 Query Match 60.9%; Score 6390.5; DB 9; Length 2005;
 Best Local Similarity 62.1%; Pred. No. 0;
 Matches 1300; Conservative 232; Mismatches 360; Indels 201; Gaps 33;
 QY 5 LPRGTSFRPRFRESLAAIEKMAEQKQAGSTTQESBEGLEPEBAPRQDLOASKKL 64
 DB 6 LVPPGDSFRPFRESLAAIEQIABEKAKRP---KQERDDEDENGPKNSDLEAGKSL 62
 QY 65 PDIYGNPPOELIGEPEDDPEFSTOKTIVLANKGTTIRFSATNALYLSPHPIRRA 124
 DB 63 PRYIGDIPPEMWSVPLEDDPYINKKTFIVANKGKAISRFSAIPALYILTPNPPIRKL 122
 QY 125 VKLIYHSLFNNLIMCTILNVCVFMAGHDPMPMKYVEYPTATYFESLVIKARGCLH 184
 DB 123 IKLIYHSLFNNLIMCTILNVCVFMAGHDPMPMKYVEYPTATYFESLVIKARGCLH 182
 QY 185 APTFLRDPNNWLDVSYIIMAYTTEFVYDLGNVSAIRTFYLRALKTISVIGLKTIVGALI 244
 DB 183 DFTFLRDPNNWLDVYITFAVYTFEYDLGNVSAIRTFYLRALKTISVIGLKTIVGALI 242
 QY 245 QSVYKCLADVMVLTVFCLSVFALIGQLFNGNLRHKVR-----NFTALNGTNGSV 294
 DB 243 QSVYKCLADVMVLTVFCLSVFALIGQLFNGNLRHKCLQPPDSSFEINITSF--FNNSL 300
 QY 295 EADGLVWE-----SIDLVLSDPENYLKNGTSDVLLCGNSSDAGCCPBGYRLKAGENP 348
 DB 301 DENGTTNRTVSIENNDEYIEDKSHFYFLEGQNDALCGNSSDAGCCPBGYRLKAGENP 360
 QY 349 DGGYTSFDSFAMAFIALFRIMTODCWERLYOQTLRSAGKIYIMFPMVYFLGFSFYVNL 408
 DB 361 NGYTSFDFSMARFSLFRIMTODPWNENYQTLRAAGTVMIFFLVYFLGFSFYVNL 420
 QY 409 LAVVAMAYEQNATTAETBEKRPQEMEMIKKEHEALTTR-----GV 453
 DB 421 LAVVAMAYEQNATTAETBEKRPQEMEMIKKEHEALTTR-----GV 480
 QY 454 DTVSRSSLEMSPLAPVNSHE---RSKRRKMSSGTEEGEDRLPKSDSEDDGR----- 504
 DB 481 GVFSSESVASKLSSSEKELNRRKKKQKQSGER--KNDRLKSESEDSIRRGCFRP 539
 QY 505 -----AMNHLSTRLGSLRTSMKPPSSRGSIPTFRRR--DLGSEADPADENS 549
 DB 540 SLEGSRLTYEKRFSSPHQSLISIRGSLSPRNRNSRSLFSFRGRADTISENDFADERS 599
 QY 550 TAGESESHRTSLVP--WPLRTSAQGPSPTSA--PGHALHGKNSYVDCNGVSLGA 606
 DB 600 TFEQDSRSDSLFVPHRDERHSNVQSASRSLVPLIPNMGKMSAVDCNGVSLVG- 658
 QY 607 GPPKATSPSHLRPMLLEHPDITTPSEBPGPQMLTSGQACVDFEPPGARORALSAV 666
 DB 659 GSTVLSAQQLL-----PGTTEITEI--RKRRSSYHVSMDLEDPTRORARMSIA 708
 QY 667 SVLTSLAEELSESRHKCPCKNRLAORVYIMECPMMSIKQGVKLVWMDPFDLITTC 726
 DB 709 SLTNIMEELSESRKCPCKNRYKFNAMCLIMCCKPWLVKALVNLVWMDPFDLITTC 768
 QY 727 IVLNTLFPALAEHYNNITSEFEEMLVQGNLVFTGIFTAEMTFKIIALDPYVYFOGNNIPDS 786
 DB 769 IVLNTLFPAMEMHYPTTEGSSVLTGVLFTGIFTAEMTKIIANDPYVYFOGNNIPFG 828
 QY 787 IIVLISLWELGSRKNSNLSVLSFLLVYFKLAKSWPTLTIKTIKNSVGLGNULTVL 846
 DB 828 FIVSLISLWELGSRKNSNLSVLSFLLVYFKLAKSWPTLTIKTIKNSVGLGNULTVL 888
 QY 847 AIVVIFAVVAGQLGKKNYSR--LRDSDGGLPRHMDFFAFILIFILCGEMLETWM 904
 DB 847 AIVVIFAVVAGQLGKKNYSR--LRDSDGGLPRHMDFFAFILIFILCGEMLETWM 904

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Db      889 AIIIVFAVAVGQOLRGSKYKEVCYCKI SNDCELPKRMHMDFFHSFLIVPRVLCGEIETQM 948
Qy      905 DCMEEVSGSGLCLLVPLLVAVVIGNLVVNLFLALLSSFSADNLTAPDEDERENNNIQLALA 964
Db      949 DCMEEVAGQMTCLTVFMVAVVIGNLVVNLFLALLSSFSADNLTAPDEDERENNNIQLAIVG 1008
Qy      965 RIQRGLRFYKRTTWDFCCGLRORPOKPAAL-----AAQGLPSCITPFSPPPEETE 1017
Db      1009 RMQKGIIDFVYKRIREF---IQAFAVKOKALDEIKPLELDJNNKSCIS----- 1054
Qy      1018 KVPPTKRETRFECE-----OPQGGTP-----GDPE-PVCVPIA 1050
Db      1055 -----NHTTIEIGKDLNVLKQNGTTSIGSSVEKCVVDESDYMFINNPSLTJYVPA 1108
Qy      1051 VASDTPDOBEDEENSLGTEEBSSKQESQPVSGGPAPDSRTSGVATASSEAEASAS 1110
Db      1109 VESDEP-----NLNTEPFSSSESDME-----BSKEKLTANTSSSEGST--- 1145
Qy      1111 QADWROQMAEPQAPCCGCTPEDSCSGSTADMTNTAEILBQIPDLQGVKQPEDCFTG 1170
Db      1146 -----VDIGAPAEGEQPE-----VEPEBSIE-----PEACFTED 1174
Qy      1171 CVRRCPCCAVDTTOAGKVMWRRLKTCYHIVHSEWPEFTFIIMIISSGALAFEDYTEE 1230
Db      1175 CVAKKCCQOISTEEGKGLMMLRKACYKI VEHAWETFI VFMIISSGALAFEDYIEQ 1234
Qy      1231 RKTIKVLEAYADRMFTYVFLVLEMLKMAVAGPKCYFTNACWIDFLIVDSIVSLVANTL 1290
Db      1235 RKTIKTMLEAYADKVFYIFILEMLKMAVAGFQVFTNACWIDFLIVDSIVSLVANTL 1294
Qy      1291 GPFAMGPYKSLRLRLRLRLRALSREGRVYNVALGALPSIMNTLVCLTFMIIESIM 1350
Db      1295 GSELSAIXSLRLRLRLRLRALSREGRVYNVALGALPSIMNTLVCLTFMIIESIM 1354
Qy      1351 GVALPAGKPGRCINOTEGDPLNTYTYNNKSQCESL---NLGELWYTKVAVFDVNGAG 1407
Db      1355 GVALPAGKPGRCINOTEGDPLNTYTYNNKSQCESL---NLGELWYTKVAVFDVNGAG 1411
Qy      1408 YLALLQVATPKGMNDIMYAAVDSRGYEEOPQWENLWMTYFYVFIIFISGFTLNFIGV 1467
Db      1412 YLALLQVATPKGMNDIMYAAVDSRGYEEOPQWENLWMTYFYVFIIFISGFTLNFIGV 1471
Qy      1468 IINFNQOKKKLGGODIEMTEBOKKYNNAMKLGSKKKPKRIPRLANKOGFIEDIVTQ 1527
Db      1472 IINFNQOKKKLGGODIEMTEBOKKYNNAMKLGSKKKPKRIPRLANKOGFIEDIVTQ 1531
Qy      1528 AFDVTIMFLICLMMVTMMVETDOSPCKINILAKINLFAFATGECIVLALRHYFF 1587
Db      1532 VFDISTIMFLICLMMVTMMVETDOSPCKINILAKINLFAFATGECIVLALRHYFF 1591
Qy      1588 NSMNIPEDFVVVILSVGTVLSDIIOKXFSPSLFRVIRLARIGRIILIRGAGIRTLF 1647
Db      1592 IGMNIPDFVVVILSVGTVLSDIIOKXFSPSLFRVIRLARIGRIILIRGAGIRTLF 1651
Qy      1648 ALMASLPALNIGLILFLVNFYISIFGMANFAYVKAAGIDDMFNQTPANSKLCFQIT 1707
Db      1652 ALMASLPALNIGLILFLVNFYISIFGMANFAYVKAAGIDDMFNQTPANSKLCFQIT 1711
Qy      1708 TSAQMDGLSPILNTGPGPYCDPTLPNSNGS-RGDCGSPAYGILFEFTYIILSVLVNMY 1766
Db      1712 TSAQMDGLSPILNTGPGPYCDPTLPNSNGS-RGDCGSPAYGILFEFTYIILSVLVNMY 1771
Qy      1767 IAILLENFSVATEBESTEPLSEDDFMEFYIWEKEFPBPATOFIEYSVLSDPADALSEPLRI 1826
Db      1772 IAILLENFSVATEBESTEPLSEDDFMEFYIWEKEFPBPATOFIEYSVLSDPADALSEPLRI 1831
Qy      1827 AKENQISLIMNDLPMSVSGDRICHMDILPATFKRYLGSSGMDALAKIQEMEKPAANPSKI 1886
Db      1832 AKENQISLIMNDLPMSVSGDRICHMDILPATFKRYLGSSGMDALAKIQEMEKPAANPSKI 1891
Qy      1887 SYERTITTLRKHEEVANVYTORAFRHLIORSJKAHSLFPROQAGSGLSEEDAPEREGL 1946
Db      1892 SYERTITTLRKHEEVANVYTORAFRHLIORSJKAHSLFPROQAGSGLSEEDAPEREGL 1949

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Qy      1947 IAYVSENFSPRLPSPSSSISSTSPSPSYSTRATSDNLQVRGSDYSHSED 1999
Db      1950 AIDKLTENST---PEKDTMPTSTSPSPSYSTRATSDNLQVRGSDYSHSED 1995

RESULT 36
AAB99677
ID AAB99677 standard; protein; 2005 AA.
XX
AC AAB99677;
XX
DT 04-SEP-2001 (first entry)
XX
DE Human neonatal form of SCN2A protein sequence SEQ ID NO:36.
XX
KW Human; epilepsy; chromosome 2; SCN1A; SCN2A; SCN3A; identification;
KW diagnosis; mutation; chromosome 2q23-q31; neurological disorder;
KW anticonvulsant; neuroprotective.
XX
OS Homo sapiens.
XX
PN WO200138564-A2.
XX
PD 31-MAY-2001.
XX
PF 24-NOV-2000; 2000WO-CA001404.
XX
PR 26-NOV-1999; 99US-0167623P.
XX
PA (UWMC-) UNIV MCGILL.
XX
PI Rouleau GA, Lafreniere RG, Rochefort D, Cossette P, Ragdale D;
XX
DR WPI; 2001-355945/37.
XX
DR N-PSDB; AAB55794.
XX
PT Determining a predisposition to epilepsy and/or development of epilepsy
PT comprises determining the genotype of SCN1A, SCN2A and/or SCN3A, or a DNA
PT variant, equivalent, or mutation which shows a linkage disequilibrium.
XX
PS Disclosure; Page 131-138; 268pp; English.
XX
CC The present invention describes a method (M1) of determining an
CC individual's predisposition to epilepsy and/or development of epilepsy,
CC as well as predicting the individual's response to medication. The method
CC comprises determining the genotype of at least one gene selected from
CC SCN1A, SCN2A or SCN3A, or a DNA variant, equivalent, or mutation which
CC shows a linkage disequilibrium. SCN1A, SCN2A and SCN3A are all sodium
CC channel genes located on chromosome 2. The idiopathic generalised
CC epilepsy (IGE) gene is more specifically localised on chromosome 2q23-
CC q31. Compounds identified as modulators of the biological activity of
CC SCN1A, SCN2A or SCN3A proteins or genes, are useful for treating epilepsy
CC or other neurological disorders. They have anticonvulsant and
CC neuroprotective activities. AAB55763 to AAB56164 and AAB99674 to AAB99679
CC represent SCN1A, SCN2A, and SCN3A cDNAs, gene fragments, PCR primers,
CC oligonucleotides and proteins given in the exemplification of the present
CC invention
XX
SQ Sequence 2005 AA;

Query Match 60.9%; Score 6389.5; DB 4; Length 2005;
Best Local Similarity 62.1%; Pred. No. 0;
Matches 1300; Conservative 232; Mismatches 360; Indels 201; Gaps 33;

Qy      5 LIPRGTSFRRTRESIAIERKMAEKQARSGSTTLQESHEGUPBEAPRPODLQASKUL 64
Db      6 LVPPEGDSFRFTRESIAIERKMAEKQARSGSTTLQESHEGUPBEAPRPODLQASKUL 62
Qy      65 PDLGNPQOELIGPELIDLPYVSTOKTFTIVANKGKTIFRSBATNALYVLSFPHPRRA 124
Db      63 PFIYGDIPPEWVSVPLEDDPYINKKFTIVANKGKISRFSATPALYLTPTFNPTRKLA 122

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QY 125 VKILVHSLFNNLIMCTILINCVFMAOHDPMPMTKYVEFTALYFESTVILARGCLH 184
 DB 123 IKILVHSLFNNLIMCTILINCVFMTNSPDMTNGVETFTGITYFESLILKILARGCLE 182
 QY 185 AFTFLRDPMMWLDSEVIIMATTEFDLGNVSALRFRVLRALKTISVIGSLKTIYVALI 244
 DB 183 DFFELRDPMMWLDFTVITFAVTEFVNLGNVSALRFRVLRALKTISVIGSLKTIYVALI 242
 QY 245 QSVYKSLADVWVLPVFCISVPAITGLQIFMGNLRKRCYR-----NPLANGTNGSV 294
 DB 243 QSVYKSLADVWVLPVFCISVPAITGLQIFMGNLRKRCYR-----NPLANGTNGSV 300
 QY 295 EADGLVME-----SLDYLSDPENYLLKNGTSDVLLCGNSSDAGCPREGYCLAKAGENP 348
 DB 301 DNGCTTNRKTVSIFNMDEYIEDKSHFYFLBQNDALICGNSSDAGCPREGYCLAKAGENP 360
 QY 349 DHGYSFDSFAMAFALFRMTODCWERLYOQTLRSAGIKYMFMLVIFLGSFYLVNLI 408
 DB 361 NGYTSFDTSMALSLFRMTODFMENTLYQTLRAAGIKYMFVFLVIFLGSFYLVNLI 420
 QY 409 LAVVAMAYEBONQATIAETEKEKRFQBAEMMLKKEHEALTIR-----GV 453
 DB 421 LAVVAMAYEBONQATIAEAOKEAFQOMLEOLKQOEAOAAAAAASBRDPSGAGGI 480
 QY 454 DTVSRSLSEMSPIAPVNSHE---RKRKRKMSGTEBCEGDBRLPKSDSEGP-----504
 DB 481 GVSSESSVASKISSKEKELKRRKKKKKKOKEGSEEB-KNDRYLKSSESDSIRKRCFRF 539
 QY 505 -----AMNHLSTRGLSRTSMKRSRSGSIPTFRRR-DLGSADPADEENS 549
 DB 540 SLBGRRLTYEKRFSSPHQSLSTRGSLFSPRNSRABLFSGRAKDIGSENPADDEHS 599
 QY 550 TAEBESHRSLVLP--WPLRRTSAGQBPRTSA-PGHALHCKNSVDCNNGVSLGA 606
 DB 600 TFDNDSRRDLSLFPVPHRGRRRHSNVSQASRASVLPILPMNGMHSADVONGVSLVG- 658
 QY 607 GDEEATPSGSHLRPVMLHPPTTPSEBEGGQWMLTQAPCYDGEFEGARORALSAV 666
 DB 659 GPGTILISAGQL-----PGTITTEI--RKRSSSYHVSMDLIEDTTSORASIA 708
 QY 667 SVLTSLALEBESRHKPCPCMNRLAORYLIMBCCPLWMSIKOGVCLVMDPFTDLITIMC 726
 DB 709 SILTNMBELESROKPCPCMYKFPANCLIMDCKPKLVKHLVNLVMDPFDLALITIC 768
 QY 727 IYVNTLPMALERTNMTSEBEMQOVGNLVTTGIFLTAEMTKIILADPYTFQCGMIFDS 766
 DB 769 IYVNTLPMALERTNMTSEBEMQOVGNLVTTGIFLTAEMTKIILADPYTFQCGMIFDS 828
 QY 787 IYVILSIMEGLSRMSNLVLSRFLRVRFLKAKSWPTLNTLKIIGNSVGALGNLTIVL 846
 DB 829 FIVSLSIMEGLANVEGLSVLSRFLRVRFLKAKSWPTLNTLKIIGNSVGALGNLTIVL 888
 QY 847 AIIIVFAVVGMLFGKNYS--LRDSGSLPRMHMDFFAFLIFRILCEMIETMW 904
 DB 889 AIIIVFAVVGMLFGSYKECVCKISNDCELPRMHMDFFHFLIVFLRLOCEMIETMW 948
 QY 905 DCHBVSQSLCLVFLVMTGNLVNLFLALLSFSADNLTAPDEDERNNNLQIALA 964
 DB 949 DCHBVSQSLCLVFLVMTGNLVNLFLALLSFSADNLTAPDEDERNNNLQIALA 1008
 QY 965 RIQRGLRFVYKRTWDFCCGLLRQRPKRAL-----AAGQLPSCIATPVSPPPETE 1017
 DB 1009 RMQKGLDFVYKRTWDFCCGLLRQRPKRAL-----AAGQLPSCIATPVSPPPETE 1054
 QY 1018 KVPTRKTRFEGB-----QPGQGP-----GDPB-PVCVPIA 1050
 DB 1055 -----NHTTIEIGKDLNLYLKDGNGTTSIGSSVEKVVYDESDYMSFINNPSTLVTVPIA 1108
 QY 1051 VASDTPDDDEBDENSLGTREBSSKQESQVSGGPRAPPSRWSQVSATASEAASAS 1110
 DB 1109 VGSISDE-----NLNTEBFSSSESDME-----ESKEKJLNATSSSEGST--- 1145
 QY 1111 QADMRQOMKAEPOAPGCGETPEBDSGBSGSTADMTNTAELLEQIPDLGQDVKQDEDCFTBS 1170

DB 1146a-----VDICAPABGEQPE-----VEPBESLE-----PEACTED 1174
 QY 1171 CVRRCPCCAVDTTQAPGKQWMLRKTCTYHIVHSWETFTIIPMILSSGALAEEDYLEB 1230
 DB 1175 CVRRCPCCAVDTTQAPGKQWMLRKTCTYHIVHSWETFTIIPMILSSGALAEEDYLEB 1234
 QY 1231 RKTIKYLEVADKQFVYVFLVLEMLKRVYVGFCKYFPMACMDLFDLVDSIVLVNLT 1290
 DB 1235 RKTIKYLEVADKQFVYVFLVLEMLKRVYVGFCKYFPMACMDLFDLVDSIVLVNLT 1294
 QY 1291 GFAPMGPISLRTIRALRPLRALSREGKRVVNNALVGAIPISIMNVLLVCLIFMLIFSIM 1350
 DB 1295 GYSELGAIKSLRTIRALRPLRALSREGKRVVNNALVGAIPISIMNVLLVCLIFMLIFSIM 1354
 QY 1351 GVNLPAGRCRCINOTGBDPLNNTYVNNKSQCESL--NLGELWTKYKVPNDVAG 1407
 DB 1355 GVNLPAGRCRCINOTGBDPLNNTYVNNKSQCESL--NLGELWTKYKVPNDVAG 1411
 QY 1408 YIALLOVATPKGMDIMYAUVDSRGYEBQPOWEVNYMYTYFVFIIFGSFETLNPITGV 1467
 DB 1412 YIALLOVATPKGMDIMYAUVDSRGYEBQPOWEVNYMYTYFVFIIFGSFETLNPITGV 1471
 QY 1468 IIDFNQOKKKLGGQDIFMTBOKKTYNNAMKLLGSKKQKPIPRPANKYQGFIDYTKQ 1527
 DB 1472 IIDFNQOKKKLGGQDIFMTBOKKTYNNAMKLLGSKKQKPIPRPANKYQGFIDYTKQ 1531
 QY 1528 APDVTIMFLICLMTVMVETDQDSPEKINIILAKNILPVALFTGECIVKALRHVYFT 1587
 DB 1532 VFDISIMILICLMTVMVETDQDSPEKINIILAKNILPVALFTGECIVKALRHVYFT 1591
 QY 1588 NSWNIEDPVVVLISYGVTLSDIIOKYPFSPFLFRITRLARIGRIILRGAGIRPLIF 1647
 DB 1592 IGNMIDPVVVLISYGVTLSDIIOKYPFSPFLFRITRLARIGRIILRGAGIRPLIF 1651
 QY 1648 ALMMSPLALFNIGLFLVWFYISIGMANFAVYKKEAGIDMFNFOTFANSMILCFQIT 1707
 DB 1652 ALMMSPLALFNIGLFLVWFYISIGMANFAVYKKEAGIDMFNFOTFANSMILCFQIT 1711
 QY 1708 TSAGMDGLSPILNTGPPYCDPTLPNSNGS-RDGCSPAVGLIFPTTYIIISFLVNNY 1766
 DB 1712 TSAGMDGLSPILNTGPPYCDPTLPNSNGS-RDGCSPAVGLIFPTTYIIISFLVNNY 1771
 QY 1767 IAILLENBSVATBESIEPESBDDFDMFYETIWEKFDDEAOFIYVLSDFADLSBPR 1826
 DB 1772 IAILLENBSVATBESIEPESBDDFDMFYETIWEKFDDEAOFIYVLSDFADLSBPR 1831
 QY 1827 AKNOISLIMDLPVMSGDRICHMDILFAFKRVLGESEGMALKIOMEKFAAANPSKI 1886
 DB 1832 AKNOISLIMDLPVMSGDRICHMDILFAFKRVLGESEGMALKIOMEKFAAANPSKI 1891
 QY 1887 SYEPIITTLRKHEVSANVIOAFRRHLLORSCLKASFLFRQAGSGLSBEDAPBRBGL 1946
 DB 1892 SYEPIITTLRKHEVSANVIOAFRRHLLORSCLKASFLFRQAGSGLSBEDAPBRBGL 1949
 QY 1947 IAYVSENSRPLGPPSSSISSTSPPSYDVTATRSNLTQVRGSDYHSHSD 1999
 DB 1950 IAYVSENSRPLGPPSSSISSTSPPSYDVTATRSNLTQVRGSDYHSHSD 1999
 RESULT 37
 ADY27147
 ID ADY27147 standard; protein; 2005 AA.
 AC ADY27147;
 AC XX
 DT 05-MAY-2005 (first entry)
 DX XX
 DS Human SCN2A variant R2230.
 XX SCN2A; anticonvulsant; muscular-Gen.; neuroprotective; antiarrhythmic;
 KW antidiabetic; antiparkinsonian; neuroleptic; tranquilizer;
 KW antidepressant; analgesic; nephrotoxic; antidiabetic; cytostatic;

KW	diagnostic; anxiety disorder; major depressive disorder; epilepsy;
KV	paralysis; hyperthermia; myasthenia gravis; heart arrhythmia; ataxia;
KM	migraine; Alzheimer's disease; Parkinson's disease; cystic fibrosis; pain;
KN	inflammation; polycystic kidney disease; phobia; schizophrenia;
KP	neuropathic pain; hyperglycemia; hyperinsulinemia; sodium channel;
KQ	mucin.
XX	Homo sapiens.
OS	Synthetic.
FH	Key Location/Qualifiers
FT	Misc-difference 223 /label= R2230
FT	/note= "wild-type Arg substituted with Gln"
XX	
PX	WO2005014863-A1.
PD	17-FEB-2005.
PF	06-AUG-2004; 2004MO-AU001051.
PR	07-AUG-2003; 2003AU-00904154.
PA	(BION-) BIONOMICS LTD.
PI	Mulley JC, Harkin LA, Dibbens LM, Phillips HA, Heron SE;
PT	Berkovic SF, Scheffer IE, Davy AJ;
DR	WPI: 2005-195767/20. N-PDSB; ADY27075.
PS	Identifying subject predisposed to disorder associated with ion channel dysfunction, involves determining presence of specific mutation event in genes encoding ion channel subunits.
XX	
XX	Claim 20; SEQ ID NO 83; 347pp; English.
CC	This invention describes a novel method of identifying a subject predisposed to disorder associated with ion channel dysfunction comprising ascertaining whether at least one of the genes encoding ion channel subunits in the subject has undergone a mutation. The invention also describes 1) isolated nucleic acid molecules encoding an isolated polypeptide which is a mutant or variant ion channel subunit (including a mutant KCNQ2 subunit, where the mutation event has occurred in C terminal domain of the subunit and leads to disturbance in the calmodulin binding affinity of the subunit) and 2) an expression vector, cells, antibodies and a method for producing a non-human transgenic animal which are all used for screening of candidate pharmaceutical agents for diagnosing or treating epilepsy or a disorder associated with ion channel dysfunction. The mutation detected in the method disrupts the functioning of an assembled ion channel so as to produce an epilepsy phenotype in the subject or produces one or more disorders associated with ion channel dysfunction such as hyper- or hypo-kalemic periodic paralysis, myotonia, malignant hyperthermia, myasthenia, cardiac arrhythmias, episodic ataxia, migraine, Alzheimer's disease, Parkinson's disease, schizophrenia, anxiety, depression, phobic obsessive symptoms, neuropathic pain, inflammatory pain, chronic/acute pain, Bartter's syndrome, polycystic kidney disease, Dent's disease, hyperinsulinemic hypoglycemia of infancy cystic fibrosis, congenital stationary night blindness and total color- blindness in the subject or to produce an epilepsy phenotype when expressed in combination with one or more additional mutations or variations in the ion channel subunit genes. The products of the invention have anticonvulsant, muscular-gen., neuroprotective, antiarrhythmic, antimigraine, nootropic, antiParkinsonian, neurolaptic, tranquillizer, antidepressant, analgesic, nephrotoxic, antidiabetic and cytostatic activity. This sequence represents a fragment of the human sodium ion channel subunit SCN2A encoded by exon 6A which contains the mutation R223Q.
SO	Sequence 2005 AA;
Query Match	60.9%; Score 6387.5; DB 9; Length 2005;
Best Local Similarity	62.1%; Pred. NO. 0;

	Matches	1299;	Conservative	233;	Mismatches	360;	Indels	201;	Gaps	33;	
Qy	5	LLPRTSSFRRTRESLIAIEKMAEKOARGSTTLQESREGLPDEEAPPQDLOASKYL								64	
Db	6	LVPPEPDSFRFPRTSRSLAIEGRIAEBEAKRP---	KOEK	DD	ED	EN	GP	PN	DL	E	62
Qy	65	PDLYGNPEOEILGEPLEDDLPFYSYQKTFIVLANKGTIFREAFATNALVYLSFPHPRRA								124	
Db	63	PIFYIDIPPEWVSLEDDLPYINKKTFIVLANKKALSRSFATPALYLTLPNPRKUA								122	
Qy	125	VIIILHSLFNNMLIMCTILTNCSYMAQHDPPTKTYVEYFTAIYTFESLVKILANGFCUH								184	
Db	123	IKILVHSLFNNMLIMCTILTNCSVPMMSNPDPDTNAVEYFTGIIYTFESIIKILANGFCUE								182	
Qy	185	AFTFLRDPNNMLDFSVYIIMAYTTEFVDLGNVSALRTFRVLRALKTI SVISGLKTYGALI								244	
Db	183	DTFLRDPNNMLDFYITFAVYTFEFDLGNVSALRTFRVLRALKTISVIPGLKTYGVSI								242	
Qy	245	GSVKKLADVYLVTFCLSVFALIGLOFMGNLRHKCVR-----	NFTAL	NG	NS	GV				294	
Db	243	GSVKKLSDVMIITVCLSVFALIGLOFMGNLRNKLQMPDNSSFEIITSF--	FNN	SL						300	
Qy	295	FADGLVME-----	SLD	LV	SD	PN	Y	L	NG	SD	348
Db	301	DNGGTTTNTSVSIFNNDEYIEDSKSHFYLEGONDLALCNSSDAGCPEGVCYKAKGRP								360	
Qy	349	DHGYSFDSFAFAFLFRILMTQDCERLYOOTRSAGKIYIMFMYIFLGSFYVNL								408	
Db	361	NGYISFDIFSMAFLSLFRILMTQDPENLYOULTRAAGKTYMIFVLYIFLGSFYVNL								420	
Qy	409	LAVANAYEONATIAETBEKRPQOEMELKKHEALTR-----	-QY							453	
Db	421	LAVANAYEONATILEAEKOEKAEQOMLEQLKQOEBAQAAAAAASERDfSGAGGI								480	
Qy	454	DYVSRSLEMSPLAPNHSHE---RSEKRRKMSSGTEEGEDRLPKSDSEDCGR-----	504								
Db	481	GVFSESSVAASKLSSKSEBKLNRRKKKKQOEKSGEE--	KND	R	V	L	K	S	B	E	539
Qy	505	-----	ANM	H	S	L	T	R	G	L	549
Db	540	SLEGSRLTYEKRRSSPHQSLISIRGSLFSPRRNRGRSLSPFRGRADIGSENFADDEHS								599	
Qy	550	TAGESESHRTSLVP--WPLRRTSAOCQSPGRTSA--	PGH	A	H	R	K	N	S	T	606
Db	600	TFEDNDSRSDSLFVPRHGRERRRSHNSOASRSARVLPILPMNGKMSAYDCNVSIVG-								658	
Qy	607	GDEPARSPGHILRAPYMLNHPDRTTPSEEPGAPQMLTQAPCVGDGEPEPGARQALSAV								666	
Db	659	GPSTLTSAGQL-----	PBG	T	T	E	T	E	T	E	708
Qy	667	SVTSALELEESRHKCPRCMNLARORYLIMECCPLMISIKOGVKLVMDPDLTITWC								726	
Db	709	SILTNNMELEEBEROKRCPRWYGFANMCIIMDCCKPMLKVKHLYNLVMDPFDVLAITIC								768	
Qy	727	IVNLTLFMALEHYNMTSEFEEMLOQGNLVTTGIFTAEMTKIITADPYYYFOQGMNIFDS								786	
Db	769	IVNLTLFMAHEHYPMTEQFSSVYLVGVLVFTGIFTAEMFLKIITAMPDYYYFOGGMNIFDG								828	
Qy	787	IYVILSLMEGLSRMSLSTVLSRFLRLRVKAKKSNPTLNTLTKIIGNSGALGNLTVL								846	
Db	829	FIVLSLMEGLINVEBELSVLRSPRLRLRVKAKKSNPTLNTLTKIIGNSGALGNLTVL								888	
Qy	847	AIIVFLPAVVGMOLEGNYS--LRDSDGLLPRMHMMDFFHAFLLIFRLICGEWIEIWM								904	
Db	889	AIIVFLPAVVGMOLEFGKSYKECVCKSINDEGLRPMHMDFFHSLIVFRVLGCEWIEIWM								948	
Qy	905	DCMEVSGQSCLLVFLVWYIGNLVNLFLALLLSFSADNLTAPDEDEBANNQLALA								964	
Db	949	DCMEVAGQCTGLYFMWVWYIGNLVNLFLALLLSFSSDNLATADDENEMNLQAVG								1008	
Qy	965	RIGRLGFVYKRTWDFCCGLRQRQKRAL-----	AAQ	O	P	S	C	I	A	P	1017
Db	1009	RMQGGIDFVYRKRIREF---IQKAFVRQKQALDEIKPLEDLNNKQDSCS-----	1054								

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QY 1018 KVEPTRKTRFEBSGE-----QPGQGTG-----GDPF-PVCVPIA 1050
Db 1055 -----NHTTIEIGKOLNLYLKDGNGITSGISGSVEKVVVDESDYMSFINNPSLTVYPIA 1108
QY 1051 VASDTDDQDEBDEBNSIGTEBESKQESQPVSGGPEAPDPDSRTWSQVSATASBEAKSAS 1110
Db 1109 VGSDEFE-----NLNTEFEFSSESDME-----ESKEKLNATSSSEGST--- 1145
QY 1111 QADWROQWKAEPQAPGCGETPEDSCSGSTADMTNTALIEQIPDLGQDVXKDEDDCTEG 1170
Db 1146 -----VDIGAPAGEQPE-----VEBESISE-----PEACPTED 1174
QY 1171 CVARCCCAVDTTQAPGKVMWRRLKTCYHIVEHSMFEFTFIIMLLSSGALAFEDITYLER 1230
Db 1175 CVARFKCCOISIEEGKGLKMMNLRKTCYKIVEHNPETFTVPMILLSSGALAFEDITYREQ 1234
QY 1231 RKTIKVLEAYADKFTYFVLEMLKMAVYGPKKYFTNACWLDPLIVDSVLSVAMTL 1290
Db 1235 RKTIKTMLEAYADKFTYFVLEMLKMAVYGFOYFTNACWLDPLIVDSVLSVAMTL 1294
QY 1291 GFAMGPIKSLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRL 1350
Db 1295 GYSELDAIKSLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRL 1354
QY 1351 GNVLPAGKFGRCINTGEGDLPLNTYTYNNKSOCESL---NLGELVYTKYKVPDNYGAG 1407
Db 1355 GNVLPAGKFGHCINTYTGEM-FDVSVYNNYSECKALIESNQIAR---KNAVKVPDNYGAG 1411
QY 1408 YTALLQVATEFKGMDIMYAAVDSRGVEBQPEWENYLYTYFVIFIIFGSFPTLNLFTGV 1467
Db 1412 YTSLLQVATEFKGMDIMYAAVDSRVNELQPKYEDNLXMYLYFYFIIIFGSPFTLNLFTGV 1471
QY 1468 IIDNFQOQKKKLGGOIEMTEBOKKYNNAMKLGSKKPKQPIRPLNKOGPIFDIVTQK 1527
Db 1472 IIDNFQOQKKKFGGOIEMTEBOKKYNNAMKLGSKKPKQPIRPLNKOGPIFDIVTQK 1531
QY 1528 APDVTIMPLICLMMYMTWETDQSEKINIILAKNILFAITGECIYVLAALRHYYFT 1587
Db 1532 VPDISIMILCLMMYMTWETDQSEKINIILAKNILFAITGECIYVLAALRHYYFT 1591
QY 1588 NSWNIPDFVNVILSIYGVLSLIIQKYFSPPLFRVYRLRLRLRLRLRLRLRLRLRLRL 1647
Db 1592 IGMNIPDFVNVILSIYGMFLAELEIKYFVSPPLFRVYRLRLRLRLRLRLRLRLRLRLRL 1651
QY 1648 ALMMSLPALFNIGLLFLVMTYSIFGMANFAYVKRBAIGIDMNFQTPANSMCLFQIT 1707
Db 1652 ALMMSLPALFNIGLLFLVMTYFVAFGMNFAYVKRBAIGIDMNFQTPANSMCLFQIT 1711
QY 1708 TSAGMDGLSPILNTGPPYCDPTLPNSNGS--RGDCGSPAVGLFPTTYIIISFLVYNNY 1766
Db 1712 TSAGMDGLAPILNTPGPPCDPTLPNSNGS--RGDCGSPAVGLFPTTYIIISFLVYNNY 1771
QY 1767 IAILLENFVATEESTEPLESDDFDMFEYIWEKFEDEATQFIEVSYLSDPADLSEPLRI 1826
Db 1772 IAVILENFVATEESTEPLESDDFEMFEYWEKFEDEATQFIEFALSDPADLSEPLRI 1831
QY 1827 AAPNQISLINMDLPWVSGDRHICMDILFAFTRKVLGSEGBMDALQIOMEKEFMAANPSKI 1886
Db 1832 AAPNQIOLIAMLPWVSGDRHICMDILFAFTRKVLGSEGBMDALRIOMEERFMAANPSKY 1891
QY 1887 SYEPTITTLRRKHEEVSAMVIOAPRRHLLQSLKASFLFRQAGSGISEBAPREGL 1946
Db 1892 STEPTITTLRRKQEEVSAILIIOAPRRYLLKQKVVKSSITYKDKKEC--DQTPKEEDT 1949
QY 1947 IAYVSENFSPRLGPPSSSISSTSPSYDSVTRATSDNLQVRGSDYSHSED 1999
Db 1950 LIDKLNENGT----PEKIDMTPTSTSPSYDSVTRATSEKKEF--KDKSEKED 1995

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RESULT 38
 ABB06027
 ID ABB06027 standard; protein; 2000 AA.

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XX AC ABB06027;
XX DT 10-MAY-2002 (first entry)
XX DE Human sodium channel SCN3A protein SEQ ID NO:4.
XX KM Human; sodium channel; SCN3A; chromosome 2q24-31;
XX KM familial hypercalcaemic periodic paralysis; motor endplate disease.
XX OS Homo sapiens.
XX PN WO200196552-A1.
XX PD 20-DEC-2001.
XX PF 12-JUN-2001; 2001WO-JP004956.
XX PR 13-JUN-2000; 2000JP-00177540.
XX PR 13-JUN-2000; 2000JP-00177544.
XX PA (NISC-) JAPAN SCI & TECHNOLOGY CORP.
XX PI Kanazawa I, Goto J, Jeong S;
XX DR MPI; 2002-098066/13.
XX DR N-PSDB; ABL39690.
XX PT Human sodium channels SCN1A and SCN3A and encoded gene, useful in
XX PT studying physiological mechanism in which excitant cells participate and
XX PT causes of diseases and developing drugs for motor endplate disease.
XX PS Claim 2; Page 72-81; 88pp; Japanese.
XX CC The present invention describes human sodium channels SCN1A and SCN3A.
XX CC The present sequence represents the human sodium channel SCN3A. SCN1A and
XX CC SCN3A have been located to the human chromosome 2 long arm, positions
XX CC 2q24 and 2q24-31 respectively. The sodium channel proteins are useful in
XX CC studying the physiological mechanism in which excitant cells participate
XX CC and cause human diseases, and in developing remedies for e.g. familial
XX CC hypercalcaemic periodic paralysis of extremities and motor endplate
XX CC disease
XX SQ Sequence 2000 AA;
Query Match 60.8%; Score 6377.5; DB 5; Length 2000;
Best Local Similarity 62.7%; Pred. No. 0;
Matches 1296; Conservative 227; Mismatches 372; Indels 171; Gaps 29;
QY 5 LIPRGTSSPRRTRESLAAIEKRMLEKQARGSTTIOESREGIPEEAPRPOLDLQASKGL 64
Db 6 LVPPEPSFRLLTRESLAAIERRAAEKAKKPKKQDN---DDEKPKPNSDLEAGKYL 61
QY 65 PDLVGNPPOELIGPELEDIDPEYSTOKTFIVLNGKKTIFRFSATNALVYLSPHPIRRAA 124
Db 62 PRTYGDIPPEWSELEDDIDPEYINKTFIVWNGKALIFRFSATNALVYLTPANPRKIA 121
QY 125 VKILVHSLFENMLIMCTIILNVCYFMAQHDPPRTKYVEYTFYALTYFESILVKILARFCLH 184
Db 1224 IYILVHSLFENMLIMCTIILNVCYFMAQHDPPRTKYVEYTFYALTYFESILVKILARFCLH 181
QY 185 AFTPLRDPNNMLDESVIIMATYTEFVDIGANVALTFRYALRLKTTISVSGKITVAGALI 244
Db 182 DFTPLRDPNNMLDFSVIIMATYTEFVDIGANVALTFRYALRLKTTISVSGKITVAGALI 241
QY 245 QSVKGLADVMVLTVCCLSVFALIGQLFMGNLRHKCVR-----NFTAL----- 287
Db 242 QSVKGLSDVMILTVCCLSVFALIGQLFMGNLRHKCVR-----NFTAL----- 301
QY 288 NGTNGSVBADGLWESLDLYLSDPENYLLKNGTSDVLLCGNSSDAGTCPEGYRCLKAGEN 347
Db 302 NGTFVNVMTSTFNNWMD---YIGDSHFVYLDGOKPRLDLCGNSDAGTCPEGYRCLKAGEN 358

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DR N-PSDB; ADK81761.
XX
PT New antisense compound targeted to a nucleic acid molecule encoding
PT Nav1.3, useful for useful for treating a disease or condition associated
PT with Nav1.3, e.g. pain, seizure disorder such as childhood seizure
PT disorder, or ataxia.
XX
PS Disclosure; SEQ ID NO 9096; 417pp; English.
XX
CC The present invention relates to an antisense compound targeted to a
CC nucleic acid molecule encoding Nav1.3, where the antisense compound
CC specifically hybridizes with and inhibits the expression of Nav1.3. The
CC compound and composition are useful for treating a disease or condition
CC associated with Nav1.3, e.g. pain including but not limited to
CC neuropathic pain, post-herpetic neuralgia, chronic pain, lower back pain,
CC diabetic neuropathy, trigeminal neuropathy, arthritic pain, acute pain,
CC pain from burns, migraine headache, cluster headache, mild-to-moderate
CC headache; seizure disorder such as childhood seizure disorder, including
CC but not limited to neonatal or infantile epilepsy; or ataxia. The present
CC sequence represents human Nav1.3 protein.
XX
SQ Sequence 2000 AA;
Query Match 60.8%; Score 6377.5; DB 8; Length 2000;
Best Local Similarity 62.7%; Pred. No. 0;
Matches 1296; Conservative 227; Mismatches 372; Indels 171; Gaps 29;
QY 5 LHPRGSSRRFRFRESLAIEKMAEKQARGSTTLOESRGLPEEAPRQDLQASKL 64
DB 6 LVPGESEFRFLFRESLAIEKRAAEKAKCPKCEDN---DDEKKPKNSDLEAGKML 61
QY 65 PDLGNPPOELIGEPEDLDPEYSTQKTFVANKGTFPPRSATNALVYSPHPIRBA 124
DB 62 PRTYGIPEPMSEPEDEDLPTIYKKTIFVWKKGALFRFSAISALYITPLMPVKRIA 121
QY 125 VKLVHSLFNLIMCTITLNCVMAQDPPMTKYVEYTFATYTESLWKLARGFCLH 184
DB 122 IKLVHSLFNLIMCTITLNCVMTLSNPPDMKVNVEYTFGITYTESLWKLARGFCLH 181
QY 185 APTFLDPMNMLDPSYITINAYTEFDLGNVSLARTFRVLRALKTTISVGLKTIYVALI 244
DB 182 DPTFLDPMNMLDPSYITINAYTEFDLGNVSLARTFRVLRALKTTISVGLKTIYVALI 241
QY 245 QSVKRLADYVNLVPECLSPALIGLQLEFNGNLRHKVR-----PFTAL----- 287
DB 242 QSVKRLADYVNLVPECLSPALIGLQLEFNGNLRHKVR-----PFTAL----- 287
QY 288 NGTNGSVEADGLVWESLDLYSDPENYLLKNGTSDVLLGNSSDAGTCEGYCLKAGEN 347
DB 302 NGTNGSVEADGLVWESLDLYSDPENYLLKNGTSDVLLGNSSDAGTCEGYCLKAGEN 358
QY 348 PHGYTSDPSFAMAFIALFLMTQDCWERYQQOTRSAGKIYMIFFMLVIFLGSFYLNL 407
DB 359 PHGYTSDPSFAMAFIALFLMTQDCWERYQQOTRSAGKIYMIFFMLVIFLGSFYLNL 418
QY 408 IIAVVMAYEONQATIAETEEKERFOEAMENLKK-EHEALTRIGVDVSR----- 458
DB 419 IIAVVMAYEONQATIAETEEKERFOEAMENLKK-EHEALTRIGVDVSR----- 458
QY 459 -----SLEMSPLAPVNSHRRSKRRKMS-----SGTEECGEDRLPKSSEDPFRAMNH 509
DB 479 GELLESSEASLTKSSGAKEMNNRKRQRORHELENNNGERDSFKSSESDSVKSSFL 538
QY 510 SLTRGLSRTSKMP-----RSSGSIPTFRRR--DLGSEADPADDEN 548
DB 539 FEMDGRRLTSDKKFCFPHOSLSTRGSLFSPRNSKTSIFSFGRKADVGSEEDFADDD 598
QY 549 STAGESSEHRTSLV--WPIARTSAQGPSPGT-SAPGHALGKNSVVDGNGVSLG 605
DB 599 STFEDESRRDLSLFVHRHGERNSNVSGASMSRRVPGLPAGKNSHTVDCNGVSLVG 658
QY 606 AGDPEATSPGSHLLRPVMLEHPDITTPSEBPGPOMLTSOAPCVDFEFGAPQORALSA 665

DB 659 -GPSALTSPTGOL-----PEEGTT-TETEVKRRRLSSYOISMELBDSGROKAVSI 708
QY 666AVSVLTALPELSESRHKCPQWNRRLAQRLLIWECCPLMMSIKQGVKLVWMDPTDITLM 725
DB 709 ASLTINFMELSESRHKCPQWNRRLAQRLLIWECCPLMMSIKQGVKLVWMDPTDITLM 768
QY 726 CIYANTLPMALSHYNNTSSEFEEMLQVGNLVFTGISPTAEMTFKLIADLPYYPQOGNNIFD 785
DB 769 CIYANTLPMALSHYNNTSSEFEEMLQVGNLVFTGISPTAEMTFKLIADLPYYPQOGNNIFD 828
QY 786 STIIVLSIMELGSRMSNLVSRFPLRVFKLAKSPWLNTLKITIGNSVGLNULTLV 845
DB 829 GIIVSISIMELGSRMSNLVSRFPLRVFKLAKSPWLNTLKITIGNSVGLNULTLV 888
QY 846 LIIIVIFAVVGMQCFGRKYSE--LRDSRGLPRHNMDFPAFLIRRIIGEMIEYM 903
DB 889 LIIIVIFAVVGMQCFGRKYSE--LRDSRGLPRHNMDFPAFLIRRIIGEMIEYM 948
QY 904 WDCMEVSGSLCLVFLVAVVGNLVNLFALILSSFSADNLTPADEDEMNNTQALAL 963
DB 949 WDCMEVSGSLCLVFLVAVVGNLVNLFALILSSFSADNLTPADEDEMNNTQALAL 1008
QY 964 ARIQRLGVRKRTWDFCCGLLRQRPQKPAALAAQQLPSCIATPYSPPEPEKVPTR 1023
DB 1009 GIMQKIDIVYKNNMBE-CFOKAFRRKPVIEIHGKIDKSCMSNNTG-----IEISKEL 1061
QY 1024 KETRPGEQPGQCPGDE-----PVCVPIAASDITDQDEEDEN 1065
DB 1062 NYLRDNGTTSVGVTGSSEKXYIDENDYMSFINPSLTVTVPVIAGESDFE----- 1113
QY 1066 SLGTESESSKQESQPVSGPEAPDPSRTSQVSATASAEASASQADRQOKAEPOAP 1125
DB 1114 NINTEFEFSSELE-----ESKELKLANSS----- 1138
QY 1126 GCGETPESSCSGSGTAD--MTNFIABLEQIPDLGQDVQKPEDCFTEGCYRRCPCAVDTT 1183
DB 1139 -----SEGSTVDVLPREGQAELEPER--EDFK-PEACFTGCIKKEPFCVSTE 1185
QY 1184 QAPGKVMRLRRTCHIVHSHNFEPTFIIMILSSGALAFBDIYLEERTIKVLEYAK 1243
DB 1186 QAPGKVMRLRRTCHIVHSHNFEPTFIIMILSSGALAFBDIYLEERTIKVLEYAK 1245
QY 1244 MFTYVFLVEMLKNVAVYGFKKFTNAMCGLDFLIYDVSIVSVLVANTLGAEMGPISLRT 1303
DB 1246 MFTYVFLVEMLKNVAVYGFKKFTNAMCGLDFLIYDVSIVSVLVANTLGAEMGPISLRT 1305
QY 1304 LRALRPLRLSRFEGRRVVNVLVGAIPISIMNVLVCLIFMLIFSIQVNLPAKGRGCI 1363
DB 1306 LRALRPLRLSRFEGRRVVNVLVGAIPISIMNVLVCLIFMLIFSIQVNLPAKGRGCI 1365
QY 1364 NOTEGDLPLNTYITVNNKQCESLNLTGELYWTKRVKVPNVNAGATIALQVATPKGMDOI 1423
DB 1366 NOTEGDLPLNTYITVNNKQCESLNLTGELYWTKRVKVPNVNAGATIALQVATPKGMDOI 1422
QY 1424 MYAAVDSRGVEQPOMEVNLWYIYFVPIIFISGFTTLLFGVLIIDNNOQKKKGGOD 1483
DB 1422 MYAAVDSRGVEQPOMEVNLWYIYFVPIIFISGFTTLLFGVLIIDNNOQKKKGGOD 1482
QY 1484 IMTEBOKKCYNAMKKGSKPKQKPIPRELNTYOGTIPDIYVTKQAFDVTIMEPLCLMNT 1543
DB 1483 IMTEBOKKCYNAMKKGSKPKQKPIPRELNTYOGTIPDIYVTKQAFDVTIMEPLCLMNT 1542
QY 1544 MNAVEDDQSPKINILAKINLFLVAFITGECIVKLAALRHYYFTNSMNIFFRVVYVLSIV 1603
DB 1543 MNAVEDDQSPKINILAKINLFLVAFITGECIVKLAALRHYYFTNSMNIFFRVVYVLSIV 1602
QY 1604 GTVLSDILOKYPESPPTLFFVILARIGRLIRIRGAKGIRTLFLPMMSLPALFNGILL 1663
DB 1603 GMPLEAMLEKYSVSTLRRVIRLARIIGRLIRIRGAKGIRTLFLPMMSLPALFNGILL 1662
QY 1664 FLWMEYISFGNANAYYKWEAGIDMNFQTFANSMCLQFOITTSAGNDGLSLPINTG 1723
DB 1663 FLWMEYISFGNANAYYKWEAGIDMNFQTFANSMCLQFOITTSAGNDGLSLPINTG 1722

Db 949 WDCMEVAGQTMCLIVFMLVMVIGNLVNLPLALLLSFSSSDNLATDDNEMNNLQIAV 1008
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Db 1009 GWMQKIDIVKRMRE-CFOKAPFRKPKVIEIHGNKIDSCMSNNTG-----IETSKEL 1061
QY 1024 KETREBEGPOGQGTGDPDE-----PVCPIAAVESDTDQEEDEEN 1065
Db 1062 NYLRONGNTTSGVGTSSVEKVIDENDYMSFINNPSLTVTVPIAVGESDPE----- 1113
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Db 1139 -----SEGSTVDVVLPRGEQATEPEB--EDFK--PEACFTEGCIKKPPCQVSTE 1185
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Db 1366 NMTTGM--FDISDVNNLSDCCALG--KQARMQNVKNFDPNVGAGYLALLOVATPFKMDI 1422
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QY 1903 SAMVIOBAFRBHLIORSLKHAFLUROAGSGLSEEDAPEBGLIAYVMSNFSPRLGPP 1962
Db 1903 SAAIIOBFRBHLIORSLKHAFLUROAGSGLSEEDAPEBGLIAYVMSNFSPRLGPP 1962
QY 1963 SSSSISSTSPPSYDVTATSDNLQ 1988
Db 1957 KTDGSSSTPPPSYDVTATSDNLQ 1988

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